DELLY: structural variant discovery by integrated paired-end and split-read analysis

DOI: 10.1093/bioinformatics/bts378
Bioinformatics, 2012, 28, i333-i339.

#	Paper	IF	Citations
1522	Recurrent mutation of the ID3 gene in Burkitt lymphoma identified by integrated genome, exome and transcriptome sequencing. **2012**, 44, 1316-20		317
1521	cnvHiTSeq: integrative models for high-resolution copy number variation detection and genotyping using population sequencing data. **2012**, 13, R120		24
1520	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. **2013**, 45, 927-32		550
1519	Bellerophon: a hybrid method for detecting interchromosomal rearrangements at base pair resolution using next-generation sequencing data. **2013**, 14 Suppl 5, S6		12
1518	Personal genomes, quantitative dynamic omics and personalized medicine. **2013**, 1, 71-90		26
1517	MATE-CLEVER: Mendelian-inheritance-aware discovery and genotyping of midsize and long indels. *Bioinformatics*, **2013**, 29, 3143-50	7.2	35
1516	Barnacle: detecting and characterizing tandem duplications and fusions in transcriptome assemblies. **2013**, 14, 550		10
1515	iSVP: an integrated structural variant calling pipeline from high-throughput sequencing data. **2013**, 7 Suppl 6, S8		23
1514	The fractured genome of HeLa cells. **2013**, 14, 111		16
1513	Integrative genomic analyses reveal an androgen-driven somatic alteration landscape in early-onset prostate cancer. **2013**, 23, 159-70		259
1512	Criteria for inference of chromothripsis in cancer genomes. **2013**, 152, 1226-36		342
1511	The genomic and transcriptomic landscape of a HeLa cell line. **2013**, 3, 1213-24		269
1510	RSVSim: an R/Bioconductor package for the simulation of structural variations. *Bioinformatics*, **2013** , 29, 1679-81	7.2	61
1509	Impact of genomic structural variation in Drosophila melanogaster based on population-scale sequencing. **2013**, 23, 568-79		53
1508	Unexpected genomic variability in clinical and environmental strains of the pathogenic yeast Candida parapsilosis. **2013**, 5, 2382-92		38
1507	Comprehensive genetic analysis identifies a pathognomonic NAB2/STAT6 fusion gene, nonrandom secondary genomic imbalances, and a characteristic gene expression profile in solitary fibrous tumor. **2013**, 52, 873-86		196
1506	On the power and the systematic biases of the detection of chromosomal inversions by paired-end genome sequencing. **2013**, 8, e61292		20

1505	PeSV-Fisher: identification of somatic and non-somatic structural variants using next generation sequencing data. **2013**, 8, e63377	16
1504	Elucidating emergence and transmission of multidrug-resistant tuberculosis in treatment experienced patients by whole genome sequencing. **2013**, 8, e83012	56
1503	MATCHCLIP: locate precise breakpoints for copy number variation using CIGAR string by matching soft clipped reads. **2013**, 4, 157	12
1502	A deterministic analysis of genome integrity during neoplastic growth in Drosophila. **2014**, 9, e87090	10
1501	Looking the cow in the eye: deletion in the NID1 gene is associated with recessive inherited cataract in Romagnola cattle. **2014**, 9, e110628	19
1500	BSSV: Bayesian based somatic structural variation identification with whole genome DNA-seq data. **2014**, 2014, 3937-40	1
1499	Tangram: a comprehensive toolbox for mobile element insertion detection. **2014**, 15, 795	39
1498	Translocations at 8q24 juxtapose MYC with genes that harbor superenhancers resulting in overexpression and poor prognosis in myeloma patients. **2014**, 4, e191	114
1497	Konnector: Connecting paired-end reads using a bloom filter de Bruijn graph. **2014**,	5
1496	Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines. **2014**, 24, 1193-208	372
1495	Euchromatic transposon insertions trigger production of novel Pi- and endo-siRNAs at the target sites in the drosophila germline. **2014**, 10, e1004138	85
1494	Extensive copy-number variation of young genes across stickleback populations. **2014**, 10, e1004830	56
1493	Generation of antigenic diversity in Plasmodium falciparum by structured rearrangement of Var genes during mitosis. **2014**, 10, e1004812	114
1492	Insights into the genetic structure and diversity of 38 South Asian Indians from deep whole-genome sequencing. **2014**, 10, e1004377	34
1491	cnvCapSeq: detecting copy number variation in long-range targeted resequencing data. **2014**, 42, e158	8
1490	A screen for F1 hybrid male rescue reveals no major-effect hybrid lethality loci in the Drosophila melanogaster autosomal genome. **2014**, 4, 2451-60	4
1489	SHEAR: sample heterogeneity estimation and assembly by reference. **2014**, 15, 84	6
1488	Algorithm of detecting structural variations in DNA sequences. **2014**,	

1487	Towards accurate characterization of clonal heterogeneity based on structural variation. **2014**, 15, 299		7
1486	TE-Tracker: systematic identification of transposition events through whole-genome resequencing. **2014**, 15, 377		25
1485	TEMP: a computational method for analyzing transposable element polymorphism in populations. **2014**, 42, 6826-38		87
1484	TIDE: Inter-chromosomal translocation and insertion detection using embeddings. **2014**,		3
1483	Computational structural variation discovery in genomes: state of the art and challenges. **2014**,		
1482	Genomic evolution of Saccharomyces cerevisiae under Chinese rice wine fermentation. **2014**, 6, 2516-26		26
1481	Long insert whole genome sequencing for copy number variant and translocation detection. **2014**, 42, e8		21
1480	Genomic organization and evolution of double minutes/homogeneously staining regions with MYC amplification in human cancer. **2014**, 42, 9131-45		67
1479	TIGRA: a targeted iterative graph routing assembler for breakpoint assembly. **2014**, 24, 310-7		69
1478	Socrates: identification of genomic rearrangements in tumour genomes by re-aligning soft clipped reads. *Bioinformatics*, **2014**, 30, 1064-1072	7.2	60
1477	Genome comparison of Candida orthopsilosis clinical strains reveals the existence of hybrids between two distinct subspecies. **2014**, 6, 1069-78		77
1476	Detection of a recurrent DNAJB1-PRKACA chimeric transcript in fibrolamellar hepatocellular carcinoma. **2014**, 343, 1010-4		281
1475	Integrated genomic analysis identifies recurrent mutations and evolution patterns driving the initiation and progression of follicular lymphoma. **2014**, 46, 176-181		475
1474	Characterization of structural variants with single molecule and hybrid sequencing approaches. *Bioinformatics*, **2014**, 30, 3458-66	7.2	50
1473	Noninvasive detection of a balanced fetal translocation from maternal plasma. **2014**, 60, 1298-305		9
1472	Comprehensive characterization of complex structural variations in cancer by directly comparing genome sequence reads. **2014**, 32, 1106-12		62
1471	FACTERA: a practical method for the discovery of genomic rearrangements at breakpoint resolution. *Bioinformatics*, **2014**, 30, 3390-3	7.2	130
1470	SPAG7 is a candidate gene for the periodic fever, aphthous stomatitis, pharyngitis and adenopathy (PFAPA) syndrome. **2014**, 15, 190-4		13

1469	Identifying driver mutations in sequenced cancer genomes: computational approaches to enable precision medicine. **2014**, 6, 5		127
1468	Discovery of recurrent structural variants in nasopharyngeal carcinoma. **2014**, 24, 300-9		25
1467	Reference-free prediction of rearrangement breakpoint reads. *Bioinformatics*, **2014**, 30, 2559-67	7.2	5
1466	PBHoney: identifying genomic variants via long-read discordance and interrupted mapping. **2014**, 15, 180		105
1465	Comparative population genomics reveals the domestication history of the peach, Prunus persica, and human influences on perennial fruit crops. **2014**, 15, 415		97
1464	The genomic landscape underlying phenotypic integrity in the face of gene flow in crows. **2014**, 344, 1410-4		372
1463	Expanding the computational toolbox for mining cancer genomes. **2014**, 15, 556-70		146
1462	Resolving complex tandem repeats with long reads. *Bioinformatics*, **2014**, 30, 3491-8	7.2	45
1461	Gustaf: Detecting and correctly classifying SVs in the NGS twilight zone. *Bioinformatics*, **2014**, 30, 3484-90	7.2	31
1460	The genome landscape of the african green monkey kidney-derived vero cell line. **2014**, 21, 673-83		130
1459	Enhancer hijacking activates GFI1 family oncogenes in medulloblastoma. **2014**, 511, 428-34		377
1458	PolyTB: a genomic variation map for Mycobacterium tuberculosis. **2014**, 94, 346-54		64
1457	Identification of structural variation in mouse genomes. **2014**, 5, 192		16
1456	Open adjacencies and k-breaks: detecting simultaneous rearrangements in cancer genomes. **2014**, 15 Suppl 6, S4		5
1455	LUMPY: a probabilistic framework for structural variant discovery. **2014**, 15, R84		737
1454	A cell-based model system links chromothripsis with hyperploidy. **2015**, 11, 828		88
1453	An NGS Workflow Blueprint for DNA Sequencing Data and Its Application in Individualized Molecular Oncology. **2015**, 14, 87-107		8
1452	Leveraging long read sequencing from a single individual to provide a comprehensive resource for benchmarking variant calling methods. **2015**, 5, 14493		10

1451	Disclosing the Hidden Structure and Underlying Mutational Mechanism of a Novel Type of Duplication CNV Responsible for Hereditary Multiple Osteochondromas. **2015**, 36, 758-63		5
1450	An integrative framework for the identification of double minute chromosomes using next generation sequencing data. **2015**, 16 Suppl 2, S1		11
1449	Detection of Genomic Structural Variants from Next-Generation Sequencing Data. **2015**, 3, 92		169
1448	Wham: Identifying Structural Variants of Biological Consequence. **2015**, 11, e1004572		60
1447	Hairless Streaks in Cattle Implicate TSR2 in Early Hair Follicle Formation. **2015**, 11, e1005427		11
1446	The genomic landscape of fibrolamellar hepatocellular carcinoma: whole genome sequencing of ten patients. **2015**, 6, 755-70		43
1445	Structural variation discovery in the cancer genome using next generation sequencing: computational solutions and perspectives. **2015**, 6, 5477-89		23
1444	Targeted massively parallel sequencing of angiosarcomas reveals frequent activation of the mitogen activated protein kinase pathway. **2015**, 6, 36041-52		71
1443	MetaSV: an accurate and integrative structural-variant caller for next generation sequencing. *Bioinformatics*, **2015**, 31, 2741-4	7.2	95
1442	CTCF/cohesin-binding sites are frequently mutated in cancer. **2015**, 47, 818-21		286
1441	Analysis of deletion breakpoints from 1,092 humans reveals details of mutation mechanisms. **2015**, 6, 7256		56
1440	Genomic landscape of rat strain and substrain variation. **2015**, 16, 357		66
1439	LargeDEL: A tool for identifying large deletions in the whole genome sequencing data. **2015**,		
1438	Global diversity lines - a five-continent reference panel of sequenced Drosophila melanogaster strains. **2015**, 5, 593-603		79
1437	BreakSeek: a breakpoint-based algorithm for full spectral range INDEL detection. **2015**, 43, 6701-13		13
1436	ScanIndel: a hybrid framework for indel detection via gapped alignment, split reads and de novo assembly. **2015**, 7, 127		29
1435	Translocational renal cell carcinoma (t(6;11)(p21;q12) with transcription factor EB (TFEB) amplification and an integrated precision approach: a case report. **2015**, 9, 281		10
1434	Dynamics of copy number variation in host races of the pea aphid. **2015**, 32, 63-80		34

1433	RAPTR-SV: a hybrid method for the detection of structural variants. *Bioinformatics*, **2015**, 31, 2084-90	7.2	14
1432	The genomic and phenotypic diversity of Schizosaccharomyces pombe. **2015**, 47, 235-41		111
1431	Long-term culture of genome-stable bipotent stem cells from adult human liver. **2015**, 160, 299-312		830
1430	A t(3;9)(q25.1;q34.3) translocation leading to OLFM1 fusion transcripts in Gilles de la Tourette syndrome, OCD and ADHD. **2015**, 225, 268-75		12
1429	Large multiallelic copy number variations in humans. **2015**, 47, 296-303		256
1428	Methods for the detection and assembly of novel sequence in high-throughput sequencing data. *Bioinformatics*, **2015**, 31, 1904-12	7.2	12
1427	Translocation Detection Using Next-Generation Sequencing. **2015**, 151-164		3
1426	Understanding and Using Information about Cancer Genomes. **2015**, 357-368.e3		
1425	Genomics and drug profiling of fatal TCF3-HLF-positive acute lymphoblastic leukemia identifies recurrent mutation patterns and therapeutic options. **2015**, 47, 1020-1029		127
1424	FermiKit: assembly-based variant calling for Illumina resequencing data. *Bioinformatics*, **2015**, 31, 3694-67.2		57
1423	Identification of copy number variants in whole-genome data using Reference Coverage Profiles. **2015**, 6, 45		15
1422	Relapsed neuroblastomas show frequent RAS-MAPK pathway mutations. **2015**, 47, 864-71		313
1421	Transposon Insertions, Structural Variations, and SNPs Contribute to the Evolution of the Melon Genome. **2015**, 32, 2760-74		50
1420	SoloDel: a probabilistic model for detecting low-frequent somatic deletions from unmatched sequencing data. *Bioinformatics*, **2015**, 31, 3105-13	7.2	3
1419	Century-scale methylome stability in a recently diverged Arabidopsis thaliana lineage. **2015**, 11, e1004920		104
1418	Assembly and diploid architecture of an individual human genome via single-molecule technologies. **2015**, 12, 780-6		383
1417	High-throughput sequencing in mutation detection: A new generation of genotoxicity tests?. **2015**, 776, 136-43		26
1416	APOBEC family mutational signatures are associated with poor prognosis translocations in multiple myeloma. **2015**, 6, 6997		176

1415	Whole-genome CNV analysis: advances in computational approaches. **2015**, 6, 138	100
1414	A decade of structural variants: description, history and methods to detect structural variation. **2015**, 14, 305-14	59
1413	A Phylogenetic Analysis of 34 Chloroplast Genomes Elucidates the Relationships between Wild and Domestic Species within the Genus Citrus. **2015**, 32, 2015-35	188
1412	SCNVSim: somatic copy number variation and structure variation simulator. **2015**, 16, 66	21
1411	Epidermolysis bullosa in Danish Hereford calves is caused by a deletion in LAMC2 gene. **2015**, 11, 23	8
1410	Whole-genome analysis of Fusarium graminearum insertional mutants identifies virulence associated genes and unmasks untagged chromosomal deletions. **2015**, 16, 261	11
1409	Characterization of 26 deletion CNVs reveals the frequent occurrence of micro-mutations within the breakpoint-flanking regions and frequent repair of double-strand breaks by templated insertions derived from remote genomic regions. **2015**, 134, 589-603	22
1408	A review of study designs and statistical methods for genomic epidemiology studies using next generation sequencing. **2015**, 6, 149	37
1407	Memorial Sloan Kettering-Integrated Mutation Profiling of Actionable Cancer Targets (MSK-IMPACT): A Hybridization Capture-Based Next-Generation Sequencing Clinical Assay for Solid Tumor Molecular Oncology. **2015**, 17, 251-64	1066
1406	Comparison against 186 canid whole-genome sequences reveals survival strategies of an ancient clonally transmissible canine tumor. **2015**, 25, 1646-55	48
1405	QuickNGS elevates Next-Generation Sequencing data analysis to a new level of automation. **2015**, 16, 487	56
1404	Assessing structural variation in a personal genome-towards a human reference diploid genome. **2015**, 16, 286	117
1403	MMBIRFinder: A Tool to Detect Microhomology-Mediated Break-Induced Replication. **2015**, 12, 799-806	6
1402	Genetic variation and the de novo assembly of human genomes. **2015**, 16, 627-40	242
1401	Alternative transcription initiation leads to expression of a novel ALK isoform in cancer. **2015**, 526, 453-7	144
1400	SV-AUTOPILOT: optimized, automated construction of structural variation discovery and benchmarking pipelines. **2015**, 16, 238	3
1399	Local sequence assembly reveals a high-resolution profile of somatic structural variations in 97 cancer genomes. **2015**, 43, 8146-56	15
1398	Genomic Characterization of Upper Tract Urothelial Carcinoma. **2015**, 68, 970-7	147

1397	Mutational Spectrum, Copy Number Changes, and Outcome: Results of a Sequencing Study of Patients With Newly Diagnosed Myeloma. **2015**, 33, 3911-20		348
1396	Population-based structural variation discovery with Hydra-Multi. *Bioinformatics*, **2015**, 31, 1286-9	7.2	15
1395	Modeling Human Severe Combined Immunodeficiency and Correction by CRISPR/Cas9-Enhanced Gene Targeting. **2015**, 12, 1668-77		71
1394	Stable recombination hotspots in birds. **2015**, 350, 928-32		187
1393	Making the difference: integrating structural variation detection tools. **2015**, 16, 852-64		36
1392	Ulysses: accurate detection of low-frequency structural variations in large insert-size sequencing libraries. *Bioinformatics*, **2015**, 31, 801-8	7.2	12
1391	Insights into structural variations and genome rearrangements in prokaryotic genomes. *Bioinformatics*, **2015**, 31, 1-9	7.2	40
1390	A genome-wide approach for detecting novel insertion-deletion variants of mid-range size. **2016**, 44, e126		8
1389	An Intronic MBTPS2 Variant Results in a Splicing Defect in Horses with Brindle Coat Texture. **2016**, 6, 2963-70		6
1388	Similar Mutation Rates but Highly Diverse Mutation Spectra in Ascomycete and Basidiomycete Yeasts. **2016**, 8, 3815-3821		26
1387	The genetic basis for ecological adaptation of the Atlantic herring revealed by genome sequencing. **2016**, 5,		103
1386	SRBreak: A Read-Depth and Split-Read Framework to Identify Breakpoints of Different Events Inside Simple Copy-Number Variable Regions. **2016**, 7, 160		6
1385	Resolving complex structural genomic rearrangements using a randomized approach. **2016**, 17, 126		24
1384	Chromosomal Translocations in the Parasite Leishmania by a MRE11/RAD50-Independent Microhomology-Mediated End Joining Mechanism. **2016**, 12, e1006117		19
1383	Pathoadaptive Mutations of Escherichia coli K1 in Experimental Neonatal Systemic Infection. **2016**, 11, e0166793		6
1382	deBGA: read alignment with de Bruijn graph-based seed and extension. *Bioinformatics*, **2016**, 32, 3224-3232	7.2	54
1381	Sparse signal recovery methods for variant detection in next-generation sequencing data. **2016**,		3
1380	Identification of ZCCHC8 as fusion partner of ROS1 in a case of congenital glioblastoma multiforme with a t(6;12)(q21;q24.3). **2016**, 55, 677-87		22

1379	Comprehensive genome and epigenome characterization of CHO cells in response to evolutionary pressures and over time. **2016**, 113, 2241-53	88
1378	Truncation and constitutive activation of the androgen receptor by diverse genomic rearrangements in prostate cancer. **2016**, 7, 13668	101
1377	Concod: Accurate consensus-based approach of calling deletions from high-throughput sequencing data. **2016**,	1
1376	Efficient generation of transgenic cattle using the DNA transposon and their analysis by next-generation sequencing. **2016**, 6, 27185	16
1375	Extensive local adaptation within the chemosensory system following Drosophila melanogaster's global expansion. **2016**, 7, ncomms11855	31
1374	Extreme mutation bias and high AT content in Plasmodium falciparum. **2017**, 45, 1889-1901	58
1373	A high-precision shallow Convolutional Neural Network based strategy for the detection of Genomic Deletions. **2016**,	0
1372	The tandem duplicator phenotype as a distinct genomic configuration in cancer. **2016**, 113, E2373-82	71
1371	Biallelic BRCA2 Mutations Shape the Somatic Mutational Landscape of Aggressive Prostate Tumors. **2016**, 98, 818-829	26
1370	Evidence for the fixation of gene duplications by positive selection in Drosophila. **2016**, 26, 787-98	40
1369	InPhaDel: integrative shotgun and proximity-ligation sequencing to phase deletions with single nucleotide polymorphisms. **2016**, 44, e111	1
1368	Application of whole genome shotgun sequencing for detection and characterization of genetically modified organisms and derived products. **2016**, 408, 4595-614	34
1367	Evaluating Cancer of the Central Nervous System Through Next-Generation Sequencing of Cerebrospinal Fluid. **2016**, 34, 2404-15	208
1366	From Wet-Lab to Variations: Concordance and Speed of Bioinformatics Pipelines for Whole Genome and Whole Exome Sequencing. **2016**, 37, 1263-1271	33
1365	Allele-Specific Quantification of Structural Variations in Cancer Genomes. **2016**, 3, 21-34	33
1364	Discovering and Genotyping Twilight Zone Deletions. **2016**, 149-173	
1363	Tissue-specific mutation accumulation in human adult stem cells during life. **2016**, 538, 260-264	523
1362	Characterization of the Poplar Pan-Genome by Genome-Wide Identification of Structural Variation. **2016**, 33, 2706-19	66

1361	SRinversion: a tool for detecting short inversions by splitting and re-aligning poorly mapped and unmapped sequencing reads. *Bioinformatics*, **2016**, 32, 3559-3565	7.2	2
1360	Degeneration of aflatoxin gene clusters in Aspergillus flavus from Africa and North America. **2016**, 6, 62		46
1359	Global analysis of somatic structural genomic alterations and their impact on gene expression in diverse human cancers. **2016**, 113, 13768-13773		36
1358	Mutated Huntingtin Causes Testicular Pathology in Transgenic Minipig Boars. **2016**, 16, 245-59		19
1357	The genomic basis of circadian and circalunar timing adaptations in a midge. **2016**, 540, 69-73		64
1356	RAG2 and XLF/Cernunnos interplay reveals a novel role for the RAG complex in DNA repair. **2016**, 7, 10529		50
1355	Structural Variant Detection by Large-scale Sequencing Reveals New Evolutionary Evidence on Breed Divergence between Chinese and European Pigs. **2016**, 6, 18501		15
1354	NGS Analysis of Somatic Mutations in Cancer Genomes. **2016**, 357-372		
1353	Scan statistics on Poisson random fields with applications in genomics. **2016**, 10,		8
1352	Detecting horizontal gene transfer by mapping sequencing reads across species boundaries. *Bioinformatics*, **2016**, 32, i595-i604	7.2	14
1351	Characterizing the impact of sustained sulfadoxine/pyrimethamine use upon the Plasmodium falciparum population in Malawi. **2016**, 15, 575		28
1350	Mining structural variants of Heduo12 using paired-end reads. **2016**,		
1349	Evidence for L1-associated DNA rearrangements and negligible L1 retrotransposition in glioblastoma multiforme. **2016**, 7, 21		19
1348	Genome-Wide Estimates of Mutation Rates and Spectrum in Schizosaccharomyces pombe Indicate CpG Sites are Highly Mutagenic Despite the Absence of DNA Methylation. **2015**, 6, 149-60		34
1347	Genomic profiling of malignant phyllodes tumors reveals aberrations in FGFR1 and PI-3 kinase/RAS signaling pathways and provides insights into intratumoral heterogeneity. **2016**, 29, 1012-27		43
1346	Next-generation sequencing-based detection of germline L1-mediated transductions. **2016**, 17, 342		7
1345	On genomic repeats and reproducibility. *Bioinformatics*, **2016**, 32, 2243-7	7.2	22
1344	Complete Genome Sequence of a CTX-M-15-Producing Escherichia coli Strain from the H30Rx Subclone of Sequence Type 131 from a Patient with Recurrent Urinary Tract Infections, Closely Related to a Lethal Urosepsis Isolate from the Patient's Sister. **2016**, 4,		9

1343	Identifying micro-inversions using high-throughput sequencing reads. **2016**, 17 Suppl 1, 4		7
1342	Genomic variation in two gametocyte non-producing Plasmodium falciparum clonal lines. **2016**, 15, 229		15
1341	Mammary analog secretory carcinoma of the thyroid gland: A primary thyroid adenocarcinoma harboring ETV6-NTRK3 fusion. **2016**, 29, 985-95		62
1340	The oncocytic subtype is genetically distinct from other pancreatic intraductal papillary mucinous neoplasm subtypes. **2016**, 29, 1058-69		54
1339	Mitochondrial DNA Variation and Heteroplasmy in Monozygotic Twins Clinically Discordant for Multiple Sclerosis. **2016**, 37, 765-75		11
1338	Robust and exact structural variation detection with paired-end and soft-clipped alignments: SoftSV compared with eight algorithms. **2016**, 17, 51-62		31
1337	Ovarian Cancers Harboring Inactivating Mutations in CDK12 Display a Distinct Genomic Instability Pattern Characterized by Large Tandem Duplications. **2016**, 76, 1882-91		66
1336	SV-Bay: structural variant detection in cancer genomes using a Bayesian approach with correction for GC-content and read mappability. *Bioinformatics*, **2016**, 32, 984-92	7.2	24
1335	Identification of large-scale genomic variation in cancer genomes using in silico reference models. **2016**, 44, e5		4
1334	Germline Variants in Targeted Tumor Sequencing Using Matched Normal DNA. **2016**, 2, 104-11		198
1333	Identification of haplotypes at the Rsv4 genomic region in soybean associated with durable resistance to soybean mosaic virus. **2016**, 129, 453-68		28
1332	Joint detection of copy number variations in parent-offspring trios. *Bioinformatics*, **2016**, 32, 1130-7	7.2	8
1331	Manta: rapid detection of structural variants and indels for germline and cancer sequencing applications. *Bioinformatics*, **2016**, 32, 1220-2	7.2	695
1330	COSMOS: accurate detection of somatic structural variations through asymmetric comparison between tumor and normal samples. **2016**, 44, e78		3
1329	Structural variation detection using next-generation sequencing data: A comparative technical review. **2016**, 102, 36-49		89
1328	Sprites: detection of deletions from sequencing data by re-aligning split reads. *Bioinformatics*, **2016**, 32, 1788-96	7.2	13
1327	Engineered Nucleases and Trinucleotide Repeat Diseases. **2016**, 139-159		
1326	Active chromatin and transcription play a key role in chromosome partitioning into topologically associating domains. **2016**, 26, 70-84		225

1325	PopIns: population-scale detection of novel sequence insertions. *Bioinformatics*, **2016**, 32, 961-7	7.2	21
1324	HySA: a Hybrid Structural variant Assembly approach using next-generation and single-molecule sequencing technologies. **2017**, 27, 793-800		20
1323	Comprehensive Profiling of the Androgen Receptor in Liquid Biopsies from Castration-resistant Prostate Cancer Reveals Novel Intra-AR Structural Variation and Splice Variant Expression Patterns. **2017**, 72, 192-200		80
1322	Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast. **2017**, 8, 14061		212
1321	Genomic hallmarks of localized, non-indolent prostate cancer. **2017**, 541, 359-364		320
1320	Germline BRCA2 mutations drive prostate cancers with distinct evolutionary trajectories. **2017**, 8, 13671		128
1319	Whole-genome analysis of introgressive hybridization and characterization of the bovine legacy of Mongolian yaks. **2017**, 49, 470-475		67
1318	Recurrent GNAQ mutations in anastomosing hemangiomas. **2017**, 30, 722-727		36
1317	Deletion of 2.7 kb near HOXD3 in an Arabian horse with occipitoatlantoaxial malformation. **2017**, 48, 287-294		12
1316	Germline variants in familial pituitary tumour syndrome genes are common in young patients and families with additional endocrine tumours. **2017**, 176, 635-644		22
1315	Re-sequencing transgenic plants revealed rearrangements at T-DNA inserts, and integration of a short T-DNA fragment, but no increase of small mutations elsewhere. **2017**, 36, 493-504		26
1314	Genomic Rearrangements in Considered as Quantitative Traits. **2017**, 205, 1425-1441		15
1313	Asymmetric subgenome selection and cis-regulatory divergence during cotton domestication. **2017** , 49, 579-587		229
1312	Genomic signatures of adaptation to wine biological ageing conditions in biofilm-forming flor yeasts. **2017**, 26, 2150-2166		29
1311	Progenitor strain introduction of Mycobacterium bovis at the wildlife-livestock interface can lead to clonal expansion of the disease in a single ecosystem. **2017**, 51, 235-238		24
1310	PGBD5 promotes site-specific oncogenic mutations in human tumors. **2017**, 49, 1005-1014		40
1309	Mutational landscape of metastatic cancer revealed from prospective clinical sequencing of 10,000 patients. **2017**, 23, 703-713		1638
1308	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. *Bioinformatics*, **2017**, 33, 2032-2033	7.2	7

1307	Single-cell template strand sequencing by Strand-seq enables the characterization of individual homologs. **2017**, 12, 1151-1176		49
1306	High-density lipoprotein receptor SCARB1 is required for carotenoid coloration in birds. **2017**, 114, 5219-5224		66
1305	Next-generation sequencing as a tool for breakpoint analysis in rearrangements of the globin gene clusters. **2017**, 39 Suppl 1, 111-120		13
1304	Genomic profiling of pelvic genital type leiomyosarcoma in a woman with a germline :c.1100delC mutation and a concomitant diagnosis of metastatic invasive ductal breast carcinoma. **2017**, 3,		6
1303	The Sequences of 1504 Mutants in the Model Rice Variety Kitaake Facilitate Rapid Functional Genomic Studies. **2017**, 29, 1218-1231		80
1302	Toolkit for automated and rapid discovery of structural variants. **2017**, 129, 3-7		16
1301	Semi-automated cancer genome analysis using high-performance computing. **2017**, 38, 1325-1335		7
1300	Genomic profiling of breast secretory carcinomas reveals distinct genetics from other breast cancers and similarity to mammary analog secretory carcinomas. **2017**, 30, 1086-1099		41
1299	Genomic analysis of oesophageal squamous-cell carcinoma identifies alcohol drinking-related mutation signature and genomic alterations. **2017**, 8, 15290		109
1298	Whole-genome sequencing of monozygotic twins discordant for schizophrenia indicates multiple genetic risk factors for schizophrenia. **2017**, 44, 295-306		30
1297	Genotyping inversions and tandem duplications. *Bioinformatics*, **2017**, 33, 4015-4023	7.2	10
1296	Genetic diversity of next generation antimalarial targets: A baseline for drug resistance surveillance programmes. **2017**, 7, 174-180		10
1295	Enhancing knowledge discovery from cancer genomics data with Galaxy. **2017**, 6, 1-13		6
1294	Microevolution of Serial Clinical Isolates of var. and. **2017**, 8,		44
1293	The comparative landscape of duplications in Heliconius melpomene and Heliconius cydno. **2017**, 118, 78-87		10
1292	Loci associated with skin pigmentation identified in African populations. **2017**, 358,		179
1291	Lightning-fast genome variant detection with GROM. **2017**, 6, 1-7		9
1290	Accurate and comprehensive analysis of single nucleotide variants and large deletions of the human mitochondrial genome in DNA and single cells. **2017**, 25, 1229-1236		9

1289	Proteogenomic Investigation of Strain Variation in Clinical Mycobacterium tuberculosis Isolates. **2017**, 16, 3841-3851	15
1288	Genomic Investigation of Balanced Chromosomal Rearrangements in Patients with Abnormal Phenotypes. **2017**, 8, 187-194	4
1287	Rapid Discovery of De Novo Deleterious Mutations in Cattle Enhances the Value of Livestock as Model Species. **2017**, 7, 11466	36
1286	Mitochondrial mutations drive prostate cancer aggression. **2017**, 8, 656	66
1285	Genome-wide reconstruction of complex structural variants using read clouds. **2017**, 14, 915-920	65
1284	The whole-genome landscape of medulloblastoma subtypes. **2017**, 547, 311-317	472
1283	Bioinformatics Data Analysis of Next-Generation Sequencing Data from Heterogeneous Tumor Samples. **2017**, 1633, 185-192	0
1282	Genetic differences between willow warbler migratory phenotypes are few and cluster in large haplotype blocks. **2017**, 1, 155-168	46
1281	Integrative whole-genome sequence analysis reveals roles of regulatory mutations in BCL6 and BCL2 in follicular lymphoma. **2017**, 7, 7040	12
1280	Catastrophic Unbalanced Genome Rearrangements Cause Somatic Loss of Berry Color in Grapevine. **2017**, 175, 786-801	53
1279	Genomic Analysis of Pigmented Epithelioid Melanocytomas Reveals Recurrent Alterations in PRKAR1A, and PRKCA Genes. **2017**, 41, 1333-1346	59
1278	Pancreatic intraductal tubulopapillary neoplasm is genetically distinct from intraductal papillary mucinous neoplasm and ductal adenocarcinoma. **2017**, 30, 1760-1772	39
1277	Genetic insights into juvenile idiopathic arthritis derived from deep whole genome sequencing. **2017**, 7, 2657	8
1276	Homologous Recombination Deficiency and Platinum-Based Therapy Outcomes in Advanced Breast Cancer. **2017**, 23, 7521-7530	82
1275	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. **2017**,	1
1274	GRIDSS: sensitive and specific genomic rearrangement detection using positional de Bruijn graph assembly. **2017**, 27, 2050-2060	127
1273	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. **2017**, 8, 1326	191
1272	Comparing sequencing assays and human-machine analyses in actionable genomics for glioblastoma. **2017**, 3, e164	27

1271	Genomics of Parallel Experimental Evolution in Drosophila. **2017**, 34, 831-842		45
1270	Pysim-sv: a package for simulating structural variation data with GC-biases. **2017**, 18, 53		10
1269	Hi-C as a tool for precise detection and characterisation of chromosomal rearrangements and copy number variation in human tumours. **2017**, 18, 125		96
1268	Discovery of large genomic inversions using long range information. **2017**, 18, 65		14
1267	Variability among Cucurbitaceae species (melon, cucumber and watermelon) in a genomic region containing a cluster of NBS-LRR genes. **2017**, 18, 138		12
1266	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. **2017**, 18, 403		9
1265	Comprehensive detection of germline variants by MSK-IMPACT, a clinical diagnostic platform for solid tumor molecular oncology and concurrent cancer predisposition testing. **2017**, 10, 33		64
1264	Genome-wide copy number variation in the bovine genome detected using low coverage sequence of popular beef breeds. **2017**, 48, 141-150		22
1263	PSE-HMM: genome-wide CNV detection from NGS data using an HMM with Position-Specific Emission probabilities. **2016**, 18, 30		0
1262	Identification of complex genomic rearrangements in cancers using CouGaR. **2017**, 27, 107-117		21
1261	Genomic profiling of malignant peritoneal mesothelioma reveals recurrent alterations in epigenetic regulatory genes BAP1, SETD2, and DDX3X. **2017**, 30, 246-254		63
1260	Genome-wide genetic variation discovery in Chinese Taihu pig breeds using next generation sequencing. **2017**, 48, 38-47		15
1259	Population genomics identifies the origin and signatures of selection of Korean weedy rice. **2017**, 15, 357-366		32
1258	PSSV: a novel pattern-based probabilistic approach for somatic structural variation identification. *Bioinformatics*, **2017**, 33, 177-183	7.2	2
1257	novoBreak: local assembly for breakpoint detection in cancer genomes. **2017**, 14, 65-67		67
1256	Pan-cancer analysis of somatic copy-number alterations implicates IRS4 and IGF2 in enhancer hijacking. **2017**, 49, 65-74		220
1255	Seeksv: an accurate tool for somatic structural variation and virus integration detection. *Bioinformatics*, **2017**, 33, 184-191	7.2	49
1254	A recurrence-based approach for validating structural variation using long-read sequencing technology. **2017**, 6, 1-9		13

1253	An Efficient Algorithm for Identifying Genomic Structural Inversion with Wide-spectrum of Length. **2017**,	
1252	Informatics for cancer immunotherapy. **2017**, 28, xii56-xii73	11
1251	CNVcaller: highly efficient and widely applicable software for detecting copy number variations in large populations. **2017**, 6, 1-12	66
1250	Detecting large deletions at base pair level by combining split read and paired read data. **2017**, 18, 413	2
1249	Detecting chromosomal structural variation using jaccard distance and parallel architecture. **2017**,	2
1248	. **2017**,	0
1247	Sparse diploid spatial biosignal recovery for genomic variation detection. **2017**,	0
1246	Inversion detection using PacBio long reads. **2017**,	2
1245	Concurrent MMBIRFinder. **2017**,	
1244	XCAVATOR: accurate detection and genotyping of copy number variants from second and third generation whole-genome sequencing experiments. **2017**, 18, 747	17
1243	CNNdel: Calling Structural Variations on Low Coverage Data Based on Convolutional Neural Networks. **2017**, 2017, 6375059	2
1242	Germline and somatic mutations in cortical malformations: Molecular defects in Argentinean patients with neuronal migration disorders. **2017**, 12, e0185103	14
1241	Genome-wide analysis of structural variants reveals genetic differences in Chinese pigs. **2017**, 12, e0186721	5
1240	Analysis of large versus small dogs reveals three genes on the canine X chromosome associated with body weight, muscling and back fat thickness. **2017**, 13, e1006661	27
1239	Whole-genome analysis of papillary kidney cancer finds significant noncoding alterations. **2017**, 13, e1006685	26
1238	Identification of copy number variation in French dairy and beef breeds using next-generation sequencing. **2017**, 49, 77	21
1237	A de novo missense mutation of FGFR2 causes facial dysplasia syndrome in Holstein cattle. **2017**, 18, 74	7
1236	ReMixT: clone-specific genomic structure estimation in cancer. **2017**, 18, 140	18

1235	Meiotic crossovers are associated with open chromatin and enriched with Stowaway transposons in potato. **2017**, 18, 203	31
1234	CLOVE: classification of genomic fusions into structural variation events. **2017**, 18, 346	4
1233	Detection and quantification of mitochondrial DNA deletions from next-generation sequence data. **2017**, 18, 407	19
1232	Comparative Genomics of an Unusual Biogeographic Disjunction in the Cotton Tribe (Gossypieae) Yields Insights into Genome Downsizing. **2017**, 9, 3328-3344	13
1231	Plasma DNA-based molecular diagnosis, prognostication, and monitoring of patients with fusion-positive sarcomas. **2017**, 2017,	24
1230	MSeq-CNV: accurate detection of Copy Number Variation from Sequencing of Multiple samples. **2018**, 8, 4009	7
1229	Copy Number Variation/Chromosomal Aberration. **2018**, 129-135	
1228	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. **2018**, 32, 2152-2166	50
1227	HiPiler: Visual Exploration of Large Genome Interaction Matrices with Interactive Small Multiples. **2018**, 24, 522-531	21
1226	A recurrent kinase domain mutation in PRKCA defines chordoid glioma of the third ventricle. **2018**, 9, 810	42
1225	The landscape of genomic alterations across childhood cancers. **2018**, 555, 321-327	603
1224	Uncovering the genetic lesions underlying the most severe form of Hirschsprung disease by whole-genome sequencing. **2018**, 26, 818-826	14
1223	Whole genome sequencing of 91 multiplex schizophrenia families reveals increased burden of rare, exonic copy number variation in schizophrenia probands and genetic heterogeneity. **2018**, 197, 337-345	10
1222	EWSR1-NFATC2 gene fusion in a soft tissue tumor with epithelioid round cell morphology and abundant stroma: a case report and review of the literature. **2018**, 81, 281-290	16
1221	Genomic diversity in ochratoxigenic and non ochratoxigenic strains of Aspergillus carbonarius. **2018** , 8, 5439	11
1220	Utility of pooled sequencing for association mapping in nonmodel organisms. **2018**, 18, 825-837	29
1219	Very short DNA segments can be detected and handled by the repair machinery during germline chromothriptic chromosome reassembly. **2018**, 39, 709-716	15
1218	Whole-exome mutational and transcriptional landscapes of combined hepatocellular cholangiocarcinoma and intrahepatic cholangiocarcinoma reveal molecular diversity. **2018**, 1864, 2360-2368	28

1217	Whole Genome Sequencing-Based Discovery of Structural Variants in Glioblastoma. **2018**, 1741, 1-29		10
1216	Genome-wide analysis of multi- and extensively drug-resistant Mycobacterium tuberculosis. **2018**, 50, 307-316		160
1215	The Hidden Genomic and Transcriptomic Plasticity of Giant Marker Chromosomes in Cancer. **2018**, 208, 951-961		10
1214	Use of deep whole-genome sequencing data to identify structure risk variants in breast cancer susceptibility genes. **2018**, 27, 853-859		15
1213	Forward genetics screen coupled with whole-genome resequencing identifies novel gene targets for improving heterologous enzyme production in Aspergillus niger. **2018**, 102, 1797-1807		9
1212	Identifying structural variants using linked-read sequencing data. *Bioinformatics*, **2018**, 34, 353-360	7.2	36
1211	Computational identification of micro-structural variations and their proteogenomic consequences in cancer. *Bioinformatics*, **2018**, 34, 1672-1681	7.2	4
1210	Detecting Somatic Mutations in Normal Cells. **2018**, 34, 545-557		53
1209	FusorSV: an algorithm for optimally combining data from multiple structural variation detection methods. **2018**, 19, 38		28
1208	SQUID: transcriptomic structural variation detection from RNA-seq. **2018**, 19, 52		14
1207	Generic accelerated sequence alignment in SeqAn using vectorization and multi-threading. *Bioinformatics*, **2018**, 34, 3437-3445	7.2	17
1206	Novel sequences, structural variations and gene presence variations of Asian cultivated rice. **2018**, 5, 180079		10
1205	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. **2018**, 50, 727-736		156
1204	Accurate detection of complex structural variations using single-molecule sequencing. **2018**, 15, 461-468		585
1203	Mutations in Mitochondrial DNA From Pancreatic Ductal Adenocarcinomas Associate With Survival Times of Patients and Accumulate as Tumors Progress. **2018**, 154, 1620-1624.e5		15
1202	Bioinformatics Analysis for Cell-Free Tumor DNA Sequencing Data. **2018**, 1754, 67-95		11
1201	SvABA: genome-wide detection of structural variants and indels by local assembly. **2018**, 28, 581-591		149
1200	Genome-wide somatic variant calling using localized colored de Bruijn graphs. **2018**, 1, 20		51

1199	Allelic decomposition and exact genotyping of highly polymorphic and structurally variant genes. **2018**, 9, 828	31
1198	Laboratory evolution reveals regulatory and metabolic trade-offs of glycerol utilization in Saccharomyces cerevisiae. **2018**, 47, 73-82	33
1197	Finding small somatic structural variants in exome sequencing data: a machine learning approach. **2018**, 33, 1145-1158	1
1196	Diagnostic application of a capture based NGS test for the concurrent detection of variants in sequence and copy number as well as LOH. **2018**, 93, 545-556	10
1195	Whole-genome sequencing of chronic lymphocytic leukaemia reveals distinct differences in the mutational landscape between IgHV and IgHV subgroups. **2018**, 32, 332-342	30
1194	An Ovarian Adenocarcinoma With Combined Low-grade Serous and Mesonephric Morphologies Suggests a Müllerian Origin for Some Mesonephric Carcinomas. **2018**, 37, 448-459	25
1193	Whole genome amplification and sequencing of a Daphnia resting egg. **2018**, 18, 118-127	10
1192	Atypical fibroxanthoma and pleomorphic dermal sarcoma harbor frequent NOTCH1/2 and FAT1 mutations and similar DNA copy number alteration profiles. **2018**, 31, 418-428	38
1191	Measuring mutation accumulation in single human adult stem cells by whole-genome sequencing of organoid cultures. **2018**, 13, 59-78	33
1190	Molecular characterization of metastatic pancreatic neuroendocrine tumors (PNETs) using whole-genome and transcriptome sequencing. **2018**, 4,	20
1189	Genomic structural variations lead to dysregulation of important coding and non-coding RNA species in dilated cardiomyopathy. **2018**, 10, 107-120	29
1188	Adenomatoid tumors of the male and female genital tract are defined by TRAF7 mutations that drive aberrant NF-kB pathway activation. **2018**, 31, 660-673	42
1187	Frequent GNAQ and GNA14 Mutations in Hepatic Small Vessel Neoplasm. **2018**, 42, 1201-1207	34
1186	High-resolution architecture and partner genes of rearrangements in lymphoma with DLBCL morphology. **2018**, 2, 2755-2765	38
1185	An Exome-seq Based Tool for Mapping and Selection of Candidate Genes in Maize Deletion Mutants. **2018**, 16, 439-450	3
1184	Predicting Local Inversions Using Rectangle Clustering and Representative Rectangle Prediction. **2018**,	2
1183	rCANID: read Clustering and Assembly-based Novel Insertion Detection tool. **2018**,	
1182	Aggressive embryonal rhabdomyosarcoma in a 3-month-old boy: A clinical and molecular analysis. **2018**, 35, 407-414	

1181	. **2018**,	0
1180	Copy number variation detection using partial alignment information. **2018**,	1
1179	Correcting genomic deletion calls with complex boundaries from next generation sequencing data. **2018**,	
1178	Cutaneous Non-Neural Granular Cell Tumors Harbor Recurrent ALK Gene Fusions. **2018**, 42, 1133-1142	17
1177	Negative Binomial Optimization for Biomedical Structural Variant Signal Reconstruction. **2018**,	1
1176	Structural Variant Prediction in Extended Pedigrees Through Sparse Negative Binomial Genome Signal Recovery. **2018**, 2018, 1311-1314	
1175	Genome Dynamics during Environmental Adaptation Reveal Strain-Specific Differences in Gene Copy Number Variation, Karyotype Instability, and Telomeric Amplification. **2018**, 9,	46
1174	The genomic basis of adaptation to calcareous and siliceous soils in Arabidopsis lyrata. **2018**, 27, 5088-5103	10
1173	Conditional genome engineering reveals canonical and divergent roles for the Hus1 component of the 9-1-1 complex in the maintenance of the plastic genome of Leishmania. **2018**, 46, 11835-11846	14
1172	Integrated Genomic, Epigenomic, and Expression Analyses of Ovarian Cancer Cell Lines. **2018**, 25, 2617-2633	49
1171	Combining accurate tumor genome simulation with crowdsourcing to benchmark somatic structural variant detection. **2018**, 19, 188	29
1170	Nonmutational mechanism of inheritance in the Archaeon. **2018**, 115, 12271-12276	7
1169	Identification of Genes Associated With Hirschsprung Disease, Based on Whole-Genome Sequence Analysis, and Potential Effects on Enteric Nervous System Development. **2018**, 155, 1908-1922.e5	38
1168	PDX models recapitulate the genetic and epigenetic landscape of pediatric T-cell leukemia. **2018**, 10,	20
1167	Computational Tools for Population Genomics. **2018**, 127-160	2
1166	SVEngine: an efficient and versatile simulator of genome structural variations with features of cancer clonal evolution. **2018**, 7,	10
1165	The sequencing and interpretation of the genome obtained from a Serbian individual. **2018**, 13, e0208901	2
1164	Advances in Next-Generation Sequencing Bioinformatics for Clinical Diagnostics: Taking Precision Oncology to the Next Level. **2018**, 1, 149-166	1

1163	Molecular Evolution of Early-Onset Prostate Cancer Identifies Molecular Risk Markers and Clinical Trajectories. **2018**, 34, 996-1011.e8	89
1162	Genome-Informed Targeted Therapy for Osteosarcoma. **2019**, 9, 46-63	130
1161	A non-coding region near controls head colour polymorphism in the Gouldian finch. **2018**, 285,	21
1160	Aristaless Controls Butterfly Wing Color Variation Used in Mimicry and Mate Choice. **2018**, 28, 3469-3474.e4	45
1159	Comparative Genomics of S and L Morphotypes Yield Insights into Niche Adaptation. **2018**, 8, 3915-3930	13
1158	STIM1 R304W causes muscle degeneration and impaired platelet activation in mice. **2018**, 76, 87-100	15
1157	Characterization of Nigerian breast cancer reveals prevalent homologous recombination deficiency and aggressive molecular features. **2018**, 9, 4181	45
1156	Functional and evolutionary genomic inferences in through genome and population sequencing of American and European aspen. **2018**, 115, E10970-E10978	38
1155	Aberrant expression impacts the pan-cancer genomic landscape. **2018**, 28, 1611-1620	19
1154	Global genetic diversity of var2csa in Plasmodium falciparum with implications for malaria in pregnancy and vaccine development. **2018**, 8, 15429	29
1153	SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. **2018**, 7,	20
1152	FNBtools: A Software to Identify Homozygous Lesions in Deletion Mutant Populations. **2018**, 9, 976	6
1151	TranSurVeyor: an improved database-free algorithm for finding non-reference transpositions in high-throughput sequencing data. **2018**, 46, e122	9
1150	Somatic inactivating PTPRJ mutations and dysregulated pathways identified in canine malignant melanoma by integrated comparative genomic analysis. **2018**, 14, e1007589	30
1149	Integrative detection and analysis of structural variation in cancer genomes. **2018**, 50, 1388-1398	147
1148	Precancerous neoplastic cells can move through the pancreatic ductal system. **2018**, 561, 201-205	55
1147	Rearrangement bursts generate canonical gene fusions in bone and soft tissue tumors. **2018**, 361,	72
1146	Mining for Structural Variations in Next-Generation Sequencing Data. **2018**,	

1145	Jointly aligning a group of DNA reads improves accuracy of identifying large deletions. **2018**, 46, e18	1
1144	The genomic landscape of TERT promoter wildtype-IDH wildtype glioblastoma. **2018**, 9, 2087	78
1143	Combining probabilistic alignments with read pair information improves accuracy of split-alignments. *Bioinformatics*, **2018**, 34, 3631-3637	7.2
1142	Organoid Profiling Identifies Common Responders to Chemotherapy in Pancreatic Cancer. **2018**, 8, 1112-1129	394
1141	Genomic profiling of metaplastic breast carcinomas reveals genetic heterogeneity and relationship to ductal carcinoma. **2018**, 31, 1661-1674	45
1140	Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line. **2018**, 28, 1126-1135	74
1139	Molecular Cytogenetics Guides Massively Parallel Sequencing of a Radiation-Induced Chromosome Translocation in Human Cells. **2018**, 190, 88-97	6
1138	Split-Read Indel and Structural Variant Calling Using PINDEL. **2018**, 1833, 95-105	8
1137	GeneFuse: detection and visualization of target gene fusions from DNA sequencing data. **2018**, 14, 843-848	19
1136	Fine Mapping and Identification of a Novel Phytophthora Root Rot Resistance Locus on Chromosome 2 in Soybean. **2018**, 9, 44	17
1135	Structural Variant Breakpoint Detection with novoBreak. **2018**, 1833, 129-141	3
1134	Validation of Genomic Structural Variants Through Long Sequencing Technologies. **2018**, 1833, 187-192	
1133	Detecting Small Inversions Using SRinversion. **2018**, 1833, 107-114	
1132	Whole genome and transcriptome maps of the entirely black native Korean chicken breed Yeonsan Ogye. **2018**, 7,	12
1131	npInv: accurate detection and genotyping of inversions using long read sub-alignment. **2018**, 19, 261	22
1130	A large-scale whole-genome sequencing analysis reveals highly specific genome editing by both Cas9 and Cpf1 (Cas12a) nucleases in rice. **2018**, 19, 84	155
1129	The Tandem Duplicator Phenotype Is a Prevalent Genome-Wide Cancer Configuration Driven by Distinct Gene Mutations. **2018**, 34, 197-210.e5	82
1128	Comprehensive Genetic Analysis of Follicular Thyroid Carcinoma Predicts Prognosis Independent of Histology. **2018**, 103, 2640-2650	41

1127	Recurrent loss of heterozygosity correlates with clinical outcome in pancreatic neuroendocrine cancer. **2018**, 3, 18		23
1126	Compound heterozygous TYK2 mutations underlie primary immunodeficiency with T-cell lymphopenia. **2018**, 8, 6956		18
1125	Indel detection from DNA and RNA sequencing data with transIndel. **2018**, 19, 270		14
1124	Genomics of a pediatric ovarian fibrosarcoma. Association with the DICER1 syndrome. **2018**, 8, 3252		10
1123	Whole-Genome Resequencing and Pan-Transcriptome Reconstruction Highlight the Impact of Genomic Structural Variation on Secondary Metabolite Gene Clusters in the Grapevine Esca Pathogen. **2018**, 9, 1784		14
1122	Clonal evolution analysis of paired anaplastic and well-differentiated thyroid carcinomas reveals shared common ancestor. **2018**, 57, 645-652		17
1121	Analytical Validation of Clinical Whole-Genome and Transcriptome Sequencing of Patient-Derived Tumors for Reporting Targetable Variants in Cancer. **2018**, 20, 822-835		14
1120	Large extracellular vesicles carry most of the tumour DNA circulating in prostate cancer patient plasma. **2018**, 7, 1505403		169
1119	Neural differentiation, selection and transcriptomic profiling of human neuromesodermal progenitor-like cells. **2018**, 145,		29
1118	Novel genetic polymorphisms associated with severe malaria and under selective pressure in North-eastern Tanzania. **2018**, 14, e1007172		29
1117	DNA breakpoint assay reveals a majority of gross duplications occur in tandem reducing VUS classifications in breast cancer predisposition genes. **2019**, 21, 683-693		9
1116	Whole-Genome Sequencing in Cancer. **2019**, 9,		14
1115	Next Generation Sequence Analysis. **2019**, 352-363		1
1114	Detecting and Annotating Rare Variants. **2019**, 388-399		2
1113	Recurring genomic structural variation leads to clonal instability and loss of productivity. **2019**, 116, 41-53		17
1112	Molecular Fingerprinting of Anatomically and Temporally Distinct B-Cell Lymphoma Samples by Next-Generation Sequencing to Establish Clonal Relatedness. **2019**, 143, 105-111		0
1111	MAVIS: merging, annotation, validation, and illustration of structural variants. *Bioinformatics*, **2019**, 35, 515-517	7.2	13
1110	The landscape of genomic copy number alterations in colorectal cancer and their consequences on gene expression levels and disease outcome. **2019**, 69, 48-61		17

1109	Variant calling and quality control of large-scale human genome sequencing data. **2019**, 3, 399-409	1
1108	SV-Pop: population-based structural variant analysis and visualization. **2019**, 20, 136	6
1107	TDNAscan: A Software to Identify Complete and Truncated T-DNA Insertions. **2019**, 10, 685	9
1106	Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. **2019**, 37, 1155-1162	427
1105	Kevlar: A Mapping-Free Framework for Accurate Discovery of De Novo Variants. **2019**, 18, 28-36	11
1104	Comprehensive detection of chromosomal translocations in lymphoproliferative disorders by massively parallel sequencing. **2019**, 12, 121-133	1
1103	Comprehensive Identification of Fim-Mediated Inversions in Uropathogenic Escherichia coli with Structural Variation Detection Using Relative Entropy. **2019**, 4,	1
1102	Concurrent chromothripsis events in a case of TP53 depleted acute myeloid leukemia with myelodysplasia-related changes. **2019**, 237, 63-68	2
1101	Highly rearranged chromosomes reveal uncoupling between genome topology and gene expression. **2019**, 51, 1272-1282	145
1100	Predicting Local Inversions Using Rectangle Clustering and Representative Rectangle Prediction. **2019**, 18, 316-323	2
1099	Comprehensive evaluation and characterisation of short read general-purpose structural variant calling software. **2019**, 10, 3240	90
1098	Integrated analyses of murine breast cancer models reveal critical parallels with human disease. **2019**, 10, 3261	22
1097	Removal of a Subset of Non-essential Genes Fully Attenuates a Highly Virulent Strain. **2019**, 10, 664	16
1096	Integrating informatics tools and portable sequencing technology for rapid detection of resistance to anti-tuberculous drugs. **2019**, 11, 41	95
1095	Analysis of overlapping heterozygous novel submicroscopic CNVs and FANCA-VPS9D1 fusion transcripts in a Fanconi anemia patient. **2019**, 64, 899-909	5
1094	An analysis of large structural variation in global Plasmodium falciparum isolates identifies a novel duplication of the chloroquine resistance associated gene. **2019**, 9, 8287	6
1093	Adaptive archaic introgression of copy number variants and the discovery of previously unknown human genes. **2019**, 366,	36
1092	A Single SNP Turns a Social Honey Bee (Apis mellifera) Worker into a Selfish Parasite. **2019**, 36, 516-526	16

1091	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. **2019**, 11, 68		44
1090	MoMI-G: modular multi-scale integrated genome graph browser. **2019**, 20, 548		12
1089	Hecaton: reliably detecting copy number variation in plant genomes using short read sequencing data. **2019**, 20, 818		2
1088	Genome-Wide Comparisons of Mutations Induced by Carbon-Ion Beam and Gamma-Rays Irradiation in Rice Resequencing Multiple Mutants. **2019**, 10, 1514		11
1087	The Temporal Expression of Global Regulator Protein CsrA Is Dually Regulated by ClpP During the Biphasic Life Cycle of. **2019**, 10, 2495		2
1086	simuG: a general-purpose genome simulator. *Bioinformatics*, **2019**, 35, 4442-4444	7.2	17
1085	Genomic Basis of Circannual Rhythm in the European Corn Borer Moth. **2019**, 29, 3501-3509.e5		36
1084	Comprehensive Analysis of CRISPR/Cas9-Mediated Mutagenesis in by Genome-wide Sequencing. **2019**, 20,		4
1083	PASA: Identifying More Credible Structural Variants of Hedou12. **2020**, 17, 1493-1503		
1082	The clinical and genetic characteristics of permanent neonatal diabetes (PNDM) in the state of Qatar. **2019**, 7, e00753		14
1081	Comprehensive Characterization of the Attenuated Double Auxotroph as an Alternative to H37Rv. **2019**, 10, 1922		10
1080	Retrotransposon insertions can initiate colorectal cancer and are associated with poor survival. **2019**, 10, 4022		26
1079	LONG GRAIN 1: a novel gene that regulates grain length in rice. **2019**, 39, 1		4
1078	The population genetics of structural variants in grapevine domestication. **2019**, 5, 965-979		110
1077	Evaluation of computational genotyping of structural variation for clinical diagnoses. **2019**, 8,		18
1076	Overview: an iPS cell stock at CiRA. **2019**, 39, 17		51
1075	Current and Promising Approaches to Identify Horizontal Gene Transfer Events in Metagenomes. **2019**, 11, 2750-2766		22
1074	Long-read assembly of the Chinese rhesus macaque genome and identification of ape-specific structural variants. **2019**, 10, 4233		26

1073	Detection of False-Positive Deletions from the Database of Genomic Variants. **2019**, 2019, 8420547		
1072	The whole-genome landscape of Burkitt lymphoma subtypes. **2019**, 134, 1598-1607		54
1071	The NSL complex maintains nuclear architecture stability via lamin A/C acetylation. **2019**, 21, 1248-1260		34
1070	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. *Bioinformatics*, **2020**, 36, 1267-1269	7.2	8
1069	Bioinformatics Workflows for Genomic Variant Discovery, Interpretation and Prioritization. **2019**,		2
1068	Genomic profiling of combined hepatocellular-cholangiocarcinoma reveals similar genetics to hepatocellular carcinoma. **2019**, 248, 164-178		49
1067	Integration of Genomic and Transcriptional Features in Pancreatic Cancer Reveals Increased Cell Cycle Progression in Metastases. **2019**, 35, 267-282.e7		80
1066	Arginine to Glutamine Variant in Olfactomedin Like 3 () Is a Candidate for Severe Goniodysgenesis and Glaucoma in the Border Collie Dog Breed. **2019**, 9, 943-954		9
1065	Exploring the landscape of focal amplifications in cancer using AmpliconArchitect. **2019**, 10, 392		59
1064	Targeted Genomic Profiling of Acral Melanoma. **2019**, 111, 1068-1077		55
1063	Duphold: scalable, depth-based annotation and curation of high-confidence structural variant calls. **2019**, 8,		25
1062	Clinical utility of targeted NGS panel with comprehensive bioinformatics analysis for patients with acute lymphoblastic leukemia. **2019**, 60, 3138-3145		4
1061	Resolving the Insertion Sites of Polymorphic Duplications Reveals a HERC2 Haplotype under Selection. **2019**, 11, 1679-1690		5
1060	Deficiency of nucleotide excision repair is associated with mutational signature observed in cancer. **2019**, 29, 1067-1077		37
1059	Integrative genomic analysis of matched primary and metastatic pediatric osteosarcoma. **2019**, 249, 319-331		19
1058	Genomic characterization of a well-differentiated grade 3 pancreatic neuroendocrine tumor. **2019**, 5,		8
1057	Clinical Genomic Sequencing of Pediatric and Adult Osteosarcoma Reveals Distinct Molecular Subsets with Potentially Targetable Alterations. **2019**, 25, 6346-6356		39
1056	Immunohistochemical and molecular features of cholangiolocellular carcinoma are similar to well-differentiated intrahepatic cholangiocarcinoma. **2019**, 32, 1486-1494		17

1055	Tracing Oncogene Rearrangements in the Mutational History of Lung Adenocarcinoma. **2019**, 177, 1842-1857.e21		24
1054	Comprehensive evaluation of structural variation detection algorithms for whole genome sequencing. **2019**, 20, 117		163
1053	Breakage-Fusion-Bridge Events Trigger Complex Genome Rearrangements and Amplifications in Developmentally Arrested T Cell Lymphomas. **2019**, 27, 2847-2858.e4		9
1052	Myxoid smooth muscle neoplasia of the uterus: comprehensive analysis by next-generation sequencing and nucleic acid hybridization. **2019**, 32, 1688-1697		7
1051	A missense mutation (p.Tyr452Cys) in the CAD gene compromises reproductive success in French Normande cattle. **2019**, 102, 6340-6356		6
1050	GenPipes: an open-source framework for distributed and scalable genomic analyses. **2019**, 8,		46
1049	Genomic profiling of well-differentiated hepatocellular neoplasms with diffuse glutamine synthetase staining reveals similar genetics across the adenoma to carcinoma spectrum. **2019**, 32, 1627-1636		8
1048	De novo genome assembly of the cichlid fish Astatotilapia latifasciata reveals a higher level of genomic polymorphism and genes related to B chromosomes. **2019**, 128, 81-96		7
1047	Gene Fusions Are Recurrent, Clinically Actionable Gene Rearrangements in Wild-Type Pancreatic Ductal Adenocarcinoma. **2019**, 25, 4674-4681		63
1046	Navigating the non-coding genome in heart development and Congenital Heart Disease. **2019**, 107, 11-23		7
1045	Hair of the Dog: Identification of a -Regulatory Module Predicted to Influence Canine Coat Composition. **2019**, 10,		9
1044	Multiple myeloma immunoglobulin lambda translocations portend poor prognosis. **2019**, 10, 1911		53
1043	Uncovering Missing Heritability in Rare Diseases. **2019**, 10,		18
1042	Structural variants in 3000 rice genomes. **2019**, 29, 870-880		62
1041	Whole-genome sequencing of human malignant mesothelioma tumours and cell lines. **2019**, 40, 724-734		18
1040	Whole genome sequencing and variant discovery in the ASPIRE autism spectrum disorder cohort. **2019**, 96, 199-206		11
1039	Genetic, epigenetic and genomic effects on variation of gene expression among grape varieties. **2019**, 99, 895-909		11
1038	SurVIndel: improving CNV calling from high-throughput sequencing data through statistical testing. *Bioinformatics*, **2019**,	7.2	1

1037	Sequencing and curation strategies for identifying candidate glioblastoma treatments. **2019**, 12, 56		5
1036	Next-Generation Sequencing of Uveal Melanoma for Detection of Genetic Alterations Predicting Metastasis. **2019**, 8, 18		27
1035	The Proteogenomic Landscape of Curable Prostate Cancer. **2019**, 35, 414-427.e6		97
1034	CRISPR/CAS9 targeted CAPTURE of mammalian genomic regions for characterization by NGS. **2019**, 9, 3587		14
1033	Ultra-High-Frequency Reprogramming of Individual Long-Term Hematopoietic Stem Cells Yields Low Somatic Variant Induced Pluripotent Stem Cells. **2019**, 26, 2580-2592.e7		10
1032	Targeted next generation sequencing can serve as an alternative to conventional tests in myeloid neoplasms. **2019**, 14, e0212228		8
1031	Genomic Analysis in the Age of Human Genome Sequencing. **2019**, 177, 70-84		112
1030	Origin and recent expansion of an endogenous gammaretroviral lineage in domestic and wild canids. **2019**, 16, 6		5
1029	Chromothripsis during telomere crisis is independent of NHEJ, and consistent with a replicative origin. **2019**, 29, 737-749		27
1028	Long-Read Based Novel Sequence Insertion Detection With rCANID. **2019**, 18, 343-352		3
1027	Evaluation of the performance of copy number variant prediction tools for the detection of deletions from whole genome sequencing data. **2019**, 94, 103174		7
1026	High-throughput sequencing. **2019**, 39-52		
1025	Genomic and transcriptomic changes complement each other in the pathogenesis of sporadic Burkitt lymphoma. **2019**, 10, 1459		49
1024	Discovery of tandem and interspersed segmental duplications using high-throughput sequencing. *Bioinformatics*, **2019**, 35, 3923-3930	7.2	17
1023	Assessment of Cas12a-mediated gene editing efficiency in plants. **2019**, 17, 1971-1984		55
1022	Clonal evolution in myeloma: the impact of maintenance lenalidomide and depth of response on the genetics and sub-clonal structure of relapsed disease in uniformly treated newly diagnosed patients. **2019**, 104, 1440-1450		39
1021	Characterization and evolutionary dynamics of complex regions in eukaryotic genomes. **2019**, 62, 467-488		8
1020	Evaluation of Whole Genome Sequencing Data. **2019**, 1956, 321-336		2

1019	Genomic analyses of an extensive collection of wild and cultivated accessions provide new insights into peach breeding history. **2019**, 20, 36	58
1018	Structural variation and fusion detection using targeted sequencing data from circulating cell free DNA. **2019**, 47, e38	9
1017	GSDcreator: An Efficient and Comprehensive Simulator for Genarating NGS Data with Population Genetic Information. **2019**,	1
1016	. **2019**,	
1015	Benchmarking of Structural Variant Detection Tools for Microorganisms. **2019**,	
1014	Germline deletion of in familial acute lymphoblastic leukemia. **2019**, 3, 1039-1046	13
1013	Paragraph: a graph-based structural variant genotyper for short-read sequence data. **2019**, 20, 291	55
1012	A Deep Learning Approach for Detecting Copy Number Variation in Next-Generation Sequencing Data. **2019**, 9, 3575-3582	10
1011	The genomic landscape of metastatic castration-resistant prostate cancers reveals multiple distinct genotypes with potential clinical impact. **2019**, 10, 5251	66
1010	Mapping Cucumber Vein Yellowing Virus Resistance in Cucumber (L.) by Using BSA-seq Analysis. **2019**, 10, 1583	12
1009	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. **2019**, 11, 79	11
1008	Structural variant calling: the long and the short of it. **2019**, 20, 246	141
1007	Functional Enhancers Shape Extrachromosomal Oncogene Amplifications. **2019**, 179, 1330-1341.e13	87
1006	GraphTyper2 enables population-scale genotyping of structural variation using pangenome graphs. **2019**, 10, 5402	43
1005	Trio deep-sequencing does not reveal unexpected off-target and on-target mutations in Cas9-edited rhesus monkeys. **2019**, 10, 5525	15
1004	Parallel Bud Mutation Sequencing Reveals that Fruit Sugar and Acid Metabolism Potentially Influence Stress in. **2019**, 20,	3
1003	Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. **2019**, 5,	15
1002	Clinical Application of Next-Generation Sequencing-Based Panel to Wild-Type Advanced Melanoma Identifies Key Oncogenic Alterations and Therapeutic Strategies. **2020**, 19, 937-944	7

1001	DeepSV: accurate calling of genomic deletions from high-throughput sequencing data using deep convolutional neural network. **2019**, 20, 665		8
1000	The genomic diversification of grapevine clones. **2019**, 20, 972		32
999	Genomic Alteration Burden in Advanced Prostate Cancer and Therapeutic Implications. **2019**, 9, 1287		14
998	Comprehensive detection of recurring genomic abnormalities: a targeted sequencing approach for multiple myeloma. **2019**, 9, 101		22
997	LinkedSV for detection of mosaic structural variants from linked-read exome and genome sequencing data. **2019**, 10, 5585		7
996	BreakID: genomics breakpoints identification to detect gene fusion events using discordant pairs and split reads. *Bioinformatics*, **2019**, 35, 2859-2861	7.2	2
995	The role of structural genomic variants in population differentiation and ecotype formation in Timema cristinae walking sticks. **2019**, 28, 1224-1237		13
994	Identifying Genomic Variations in Monozygotic Twins Discordant for Autism Spectrum Disorder Using Whole-Genome Sequencing. **2019**, 14, 204-211		7
993	Molecular landmarks of tumor hypoxia across cancer types. **2019**, 51, 308-318		255
992	Cluster expansion of apolipoprotein D (ApoD) genes in teleost fishes. **2019**, 19, 9		1
991	Whole genomes define concordance of matched primary, xenograft, and organoid models of pancreas cancer. **2019**, 15, e1006596		29
990	Complex Structural Variant Associated with Non-syndromic Canine Retinal Degeneration. **2019**, 9, 425-437		10
989	Computational Analysis of Structural Variation in Cancer Genomes. **2019**, 1878, 65-83		0
988	CRTC1-MAML2 fusion in mucoepidermoid carcinoma of the breast. **2019**, 74, 463-473		19
987	Alfred: interactive multi-sample BAM alignment statistics, feature counting and feature annotation for long- and short-read sequencing. *Bioinformatics*, **2019**, 35, 2489-2491	7.2	29
986	Fine-Grained Analysis of Spontaneous Mutation Spectrum and Frequency in. **2019**, 211, 703-714		46
985	Sequence variants associating with urinary biomarkers. **2019**, 28, 1199-1211		13
984	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. **2019**, 21, 1611-1620		52

983	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. **2019**, 21, 1111-1120		25
982	Identification of high-risk human papillomavirus and Rb/E2F pathway genomic alterations in mutually exclusive subsets of colorectal neuroendocrine carcinoma. **2019**, 32, 290-305		26
981	DICER1 mutations are frequent in mllerian adenosarcomas and are independent of rhabdomyosarcomatous differentiation. **2019**, 32, 280-289		33
980	Bioinformatics for precision oncology. **2019**, 20, 778-788		30
979	Copy number variation detection in Chinese indigenous cattle by whole genome sequencing. **2020**, 112, 831-836		12
978	Integrated paired-end enhancer profiling and whole-genome sequencing reveals recurrent and enhancer hijacking in primary gastric adenocarcinoma. **2020**, 69, 1039-1052		21
977	A patient with pontocerebellar hypoplasia type 6: Novel RARS2 mutations, comparison to previously published patients and clinical distinction from PEHO syndrome. **2020**, 63, 103766		4
976	Meltos: multi-sample tumor phylogeny reconstruction for structural variants. *Bioinformatics*, **2020**, 36, 1082-1090	7.2	6
975	A New Next-Generation Sequencing Strategy for the Simultaneous Analysis of Mutations and Chromosomal Rearrangements at DNA Level in Acute Myeloid Leukemia Patients. **2020**, 22, 60-71		5
974	Insertional oncogenesis by HPV70 revealed by multiple genomic analyses in a clinically HPV-negative cervical cancer. **2020**, 59, 84-95		2
973	Detecting inherited and novel structural variants in low-coverage parent-child sequencing data. **2020**, 173, 61-68		0
972	A new era of long-read sequencing for cancer genomics. **2020**, 65, 3-10		28
971	Melanocytic tumors with MAP3K8 fusions: report of 33 cases with morphological-genetic correlations. **2020**, 33, 846-857		17
970	Gene duplication and transposition of mobile elements drive evolution of the Rpv3 resistance locus in grapevine. **2020**, 101, 529-542		14
969	Comparative genome-scale analysis of Pichia pastoris variants informs selection of an optimal base strain. **2020**, 117, 543-555		19
968	Structural variant identification and characterization. **2020**, 28, 31-47		6
967	Disentangling Population History and Character Evolution among Hybridizing Lineages. **2020**, 37, 1295-1305		3
966	Genetic Basis of De Novo Appearance of Carotenoid Ornamentation in Bare Parts of Canaries. **2020** , 37, 1317-1328		18

965	Genetic analysis of pleomorphic and florid lobular carcinoma in situ variants: frequent ERBB2/ERBB3 alterations and clonal relationship to classic lobular carcinoma in situ and invasive lobular carcinoma. **2020**, 33, 1078-1091	9
964	Single-cell analysis of structural variations and complex rearrangements with tri-channel processing. **2020**, 38, 343-354	17
963	Structural variation in the sequencing era. **2020**, 21, 171-189	129
962	Analysis pipelines for cancer genome sequencing in mice. **2020**, 15, 266-315	12
961	Deep sequencing of myxoinflammatory fibroblastic sarcoma. **2020**, 59, 309-317	3
960	Junction Location Identifier (JuLI): Accurate Detection of DNA Fusions in Clinical Sequencing for Precision Oncology. **2020**, 22, 304-318	1
959	Super-Pangenome by Integrating the Wild Side of a Species for Accelerated Crop Improvement. **2020**, 25, 148-158	87
958	Extrachromosomal circular DNA drives oncogenic genome remodeling in neuroblastoma. **2020**, 52, 29-34	70
957	Targeted deep-intronic sequencing in a cohort of unexplained cases of suspected Lynch syndrome. **2020**, 28, 597-608	4
956	Loss of Setd4 delays radiation-induced thymic lymphoma in mice. **2020**, 86, 102754	3
955	Spitz melanoma is a distinct subset of spitzoid melanoma. **2020**, 33, 1122-1134	31
954	Identification of gene fusion events in that encode chimeric proteins. **2020**, 2, lqaa033	1
953	A random forest-based framework for genotyping and accuracy assessment of copy number variations. **2020**, 2, lqaa071	4
952	An Increased Burden of Highly Active Retrotransposition Competent L1s Is Associated with Parkinson's Disease Risk and Progression in the PPMI Cohort. **2020**, 21,	13
951	An enhanced genetic model of relapsed IGH-translocated multiple myeloma evolutionary dynamics. **2020**, 10, 101	2
950	Reconstruction of clone- and haplotype-specific cancer genome karyotypes from bulk tumor samples. **2020**, 30, 1274-1290	5
949	Distinct Classes of Complex Structural Variation Uncovered across Thousands of Cancer Genome Graphs. **2020**, 183, 197-210.e32	45
948	An integrated personal and population-based Egyptian genome reference. **2020**, 11, 4719	6

947	Genome resequencing data for Iranian local dogs and wolves. **2020**, 13, 436	
946	Mucinous Adenocarcinoma of the Rectum: A Whole Genome Sequencing Study. **2020**, 10, 1682	3
945	An integrated peach genome structural variation map uncovers genes associated with fruit traits. **2020**, 21, 258	27
944	MYB61 is regulated by GRF4 and promotes nitrogen utilization and biomass production in rice. **2020**, 11, 5219	20
943	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. **2020**, 3, e2018109	13
942	Invasive plasmacytoid urothelial carcinoma: A comparative study of E-cadherin and P120 catenin. **2020**, 102, 54-59	2
941	Evolutionary Genomics of Structural Variation in Asian Rice (Oryza sativa) Domestication. **2020**, 37, 3507-3524	19
940	Personalised mapping of tumour development in synchronous colorectal cancer patients. **2020**, 5, 27	1
939	Identification of copy number variation and population analysis of the sacred lotus (). **2020**, 84, 2037-2044	0
938	Genomic evidence supporting the clonal expansion of extensively drug-resistant tuberculosis bacteria belonging to a rare protoBeijing genotype. **2020**, 9, 2632-2641	1
937	Highly accurate long-read HiFi sequencing data for five complex genomes. **2020**, 7, 399	39
936	Dynamic and reversible DNA methylation changes induced by genome separation and merger of polyploid wheat. **2020**, 18, 171	12
935	The evolution of relapse of adult T cell acute lymphoblastic leukemia. **2020**, 21, 284	5
934	Genomic and transcriptomic alterations associated with drug vulnerabilities and prognosis in adenocarcinoma at the gastroesophageal junction. **2020**, 11, 6091	7
933	Insertion variants missing in the human reference genome are widespread among human populations. **2020**, 18, 167	2
932	Performance of copy number variants detection based on whole-genome sequencing by DNBSEQ platforms. **2020**, 21, 518	3
931	A 50-kb deletion disrupting the RSPO2 gene is associated with tetradysmelia in Holstein Friesian cattle. **2020**, 52, 68	0
930	Predicting favorable landing pads for targeted integrations in Chinese hamster ovary cell lines by learning stability characteristics from random transgene integrations. **2020**, 18, 3632-3648	7

929	Reference Genome for the Highly Transformable ME034V. **2020**, 10, 3467-3478	13
928	Long-read-based human genomic structural variation detection with cuteSV. **2020**, 21, 189	35
927	A 63-bp insertion in exon 2 of the porcine KIF21A gene is associated with arthrogryposis multiplex congenita. **2020**, 51, 820-823	3
926	Bovine breed-specific augmented reference graphs facilitate accurate sequence read mapping and unbiased variant discovery. **2020**, 21, 184	19
925	Best practices for variant calling in clinical sequencing. **2020**, 12, 91	39
924	Complex mosaic structural variations in human fetal brains. **2020**, 30, 1695-1704	9
923	DINTD: Detection and Inference of Tandem Duplications From Short Sequencing Reads. **2020**, 11, 924	1
922	Structure and Sequence of the Sex Determining Locus in Two Wild Populations of Nile Tilapia. **2020**, 11,	6
921	VAV2 signaling promotes regenerative proliferation in both cutaneous and head and neck squamous cell carcinoma. **2020**, 11, 4788	11
920	Clinicopathological findings of pediatric NTRK fusion mesenchymal tumors. **2020**, 15, 114	7
919	ScanITD: Detecting internal tandem duplication with robust variant allele frequency estimation. **2020**, 9,	4
918	Detection of simple and complex de novo mutations with multiple reference sequences. **2020**, 30, 1154-1169	5
917	Using short read sequencing to characterise balanced reciprocal translocations in pigs. **2020**, 21, 576	3
916	Integrative genomic analysis of salivary duct carcinoma. **2020**, 10, 14995	4
915	Revealing the impact of structural variants in multiple myeloma. **2020**, 1, 258-273	28
914	Shiny-SoSV: A web-based performance calculator for somatic structural variant detection. **2020**, 15, e0238108	
913	Clinical and genomic characterisation of mismatch repair deficient pancreatic adenocarcinoma. **2021**, 70, 1894-1903	26
912	A Practical Guide for Structural Variation Detection in the Human Genome. **2020**, 107, e103	2

911	Genome-Wide Analysis of Off-Target CRISPR/Cas9 Activity in Single-Cell-Derived Human Hematopoietic Stem and Progenitor Cell Clones. **2020**, 11,	1
910	CCDC66 frameshift variant associated with a new form of early-onset progressive retinal atrophy in Portuguese Water Dogs. **2020**, 10, 21162	0
909	Identification and population genetic analyses of copy number variations in six domestic goat breeds and Bezoar ibexes using next-generation sequencing. **2020**, 21, 840	0
908	Robust barcoding and identification of Mycobacterium tuberculosis lineages for epidemiological and clinical studies. **2020**, 12, 114	24
907	Application of genome-wide insertion/deletion markers on genetic structure analysis and identity signature of Malus accessions. **2020**, 20, 540	3
906	Mutations and Copy Number Alterations in Wild-Type Glioblastomas Are Shaped by Different Oncogenic Mechanisms. **2020**, 8,	2
905	RKDOSCNV: A Local Kernel Density-Based Approach to the Detection of Copy Number Variations by Using Next-Generation Sequencing Data. **2020**, 11, 569227	2
904	Whole genome investigation of an atypical autism case identifies a novel mutation with subsequent diagnosis of Kallmann syndrome. **2020**, 23, 100593	1
903	Balancing selection in Pattern Recognition Receptor signalling pathways is associated with gene function and pleiotropy in a wild rodent. **2020**, 29, 1990-2003	3
902	Whole genome re-sequencing of sweet cherry (L.) yields insights into genomic diversity of a fruit species. **2020**, 7, 60	10
901	Genomic and Clinicopathologic Characteristics of PRKAR1A-inactivated Melanomas: Toward Genetic Distinctions of Animal-type Melanoma/Pigment Synthesizing Melanoma. **2020**, 44, 805-816	15
900	A draft genome of sweet cherry (Prunus avium L.) reveals genome-wide and local effects of domestication. **2020**, 103, 1420-1432	6
899	Activation of cryptic splicing in bovine WDR19 is associated with reduced semen quality and male fertility. **2020**, 16, e1008804	11
898	A newly developed capture-based sequencing panel for genomic assay of lung cancer. **2020**, 42, 751-759	1
897	Evolution and structure of clinically relevant gene fusions in multiple myeloma. **2020**, 11, 2666	12
896	13q12.2 deletions in acute lymphoblastic leukemia lead to upregulation of FLT3 through enhancer hijacking. **2020**, 136, 946-956	17
895	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. **2020**, 106, 872-884	36
894	Functional annotation of rare structural variation in the human brain. **2020**, 11, 2990	18

893	Recurrent inversion toggling and great ape genome evolution. **2020**, 52, 849-858	13
892	Loss of NF2 defines a genetic subgroup of non-FOS-rearranged osteoblastoma. **2020**, 6, 231-237	2
891	Detecting transcriptomic structural variants in heterogeneous contexts via the Multiple Compatible Arrangements Problem. **2020**, 15, 9	3
890	Deleterious mis-splicing of STK11 caused by a novel single-nucleotide substitution in the 3' polypyrimidine tract of intron five. **2020**, 8, e1381	1
889	Analysis of structural variants in four African cichlids highlights an association with developmental and immune related genes. **2020**, 20, 69	2
888	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. **2020**, 11, 2928	11
887	Whole-genome resequencing of wild and domestic sheep identifies genes associated with morphological and agronomic traits. **2020**, 11, 2815	48
886	Comparative genomics of high grade neuroendocrine carcinoma of the cervix. **2020**, 15, e0234505	5
885	Repeat-induced point mutation in Neurospora crassa causes the highest known mutation rate and mutational burden of any cellular life. **2020**, 21, 142	9
884	Genomics of Clinal Local Adaptation in Under Continuous Environmental and Spatial Genetic Setting. **2020**, 10, 2683-2696	7
883	Major Impacts of Widespread Structural Variation on Gene Expression and Crop Improvement in Tomato. **2020**, 182, 145-161.e23	171
882	SVXplorer: Three-tier approach to identification of structural variants via sequential recombination of discordant cluster signatures. **2020**, 16, e1007737	
881	InvBFM: finding genomic inversions from high-throughput sequence data based on feature mining. **2020**, 21, 173	1
880	Recessive missense LAMP3 variant associated with defect in lamellar body biogenesis and fatal neonatal interstitial lung disease in dogs. **2020**, 16, e1008651	3
879	Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication. **2020**, 21, 207	1
878	VALOR2: characterization of large-scale structural variants using linked-reads. **2020**, 21, 72	4
877	Mapping of the chicken cleft primary palate mutation on chromosome 11 and sequencing of the 4.9 Mb linked region. **2020**, 51, 423-429	1
876	HiNT: a computational method for detecting copy number variations and translocations from Hi-C data. **2020**, 21, 73	27

875	Germline Elongator mutations in Sonic Hedgehog medulloblastoma. **2020**, 580, 396-401	47
874	Four novel mutations of FAM20A in amelogenesis imperfecta type IG and review of literature for its genotype and phenotype spectra. **2020**, 295, 923-931	6
873	PostSV: A Post-Processing Approach for Filtering Structural Variations. **2020**, 14, 1177932219892957	0
872	NanoVar: accurate characterization of patients' genomic structural variants using low-depth nanopore sequencing. **2020**, 21, 56	27
871	Asexual Evolution and Forest Conditions Drive Genetic Parallelism in. **2020**, 8,	3
870	Molecular Diagnostics of Non-Hodgkin Lymphoma. **2020**, 26, 186-194	2
869	Pre-clinical study of induced pluripotent stem cell-derived dopaminergic progenitor cells for Parkinson's disease. **2020**, 11, 3369	93
868	Discovery and population genomics of structural variation in a songbird genus. **2020**, 11, 3403	27
867	A genome-wide survey of copy number variations reveals an asymmetric evolution of duplicated genes in rice. **2020**, 18, 73	2
866	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. **2020**, 13, 85	1
865	Primary mammary angiosarcomas harbor frequent mutations in KDR and PIK3CA and show evidence of distinct pathogenesis. **2020**, 33, 1518-1526	6
864	Wide spectrum and high frequency of genomic structural variation, including transposable elements, in large double-stranded DNA viruses. **2020**, 6, vez060	16
863	Comparative Analysis of Structural Variations Due to Genome Shuffling of Bacillus Subtilis VS15 for Improved Cellulase Production. **2020**, 21,	3
862	Identification of key genes related to seedlessness by genome-wide detection of structural variation and transcriptome analysis in 'Shijiwuhe' pear. **2020**, 738, 144480	0
861	Osteogenesis imperfecta in a male holstein calf associated with a possible oligogenic origin. **2020**, 40, 58-67	2
860	Genome sequencing identifies a homozygous inversion disrupting QDPR as a cause for dihydropteridine reductase deficiency. **2020**, 8, e1154	3
859	Transcription phenotypes of pancreatic cancer are driven by genomic events during tumor evolution. **2020**, 52, 231-240	148
858	Patterns of somatic structural variation in human cancer genomes. **2020**, 578, 112-121	232

857	Pan-cancer analysis of whole genomes. **2020**, 578, 82-93	840
856	Disruption of chromatin folding domains by somatic genomic rearrangements in human cancer. **2020**, 52, 294-305	81
855	Comparison of multiple algorithms to reliably detect structural variants in pears. **2020**, 21, 61	4
854	Varlociraptor: enhancing sensitivity and controlling false discovery rate in somatic indel discovery. **2020**, 21, 98	3
853	Review: Understanding Rare Genetic Diseases in Low Resource Regions Like Jammu and Kashmir - India. **2020**, 11, 415	7
852	Mutational signatures are jointly shaped by DNA damage and repair. **2020**, 11, 2169	70
851	Integrated immunohistochemical and molecular analysis improves diagnosis of high-grade carcinoma in the urinary bladder of patients with prior radiation therapy for prostate cancer. **2020**, 33, 1802-1810	4
850	Extrachromosomal DNA-relieving heredity constraints, accelerating tumour evolution. **2020**, 31, 884-893	31
849	Pan-cancer analysis of advanced patient tumors reveals interactions between therapy and genomic landscapes.. **2020**, 1, 452-468	34
848	diploid genome assembly for genome-wide structural variant detection. **2020**, 2, lqz018	4
847	On Variant Discovery in Genomes of Fungal Plant Pathogens. **2020**, 11, 626	9
846	Integrative multiplatform molecular profiling of benign prostatic hyperplasia identifies distinct subtypes. **2020**, 11, 1987	14
845	Genomic landscape of metastatic papillary thyroid carcinoma and novel biomarkers for predicting distant metastasis. **2020**, 111, 2163-2173	14
844	De novo somatic mutations and KRAS amplification are associated with cholangiocarcinoma in a patient with a history of choledochal cyst. **2020**, 55, 2657-2661	3
843	Increased burden of ultra-rare structural variants localizing to boundaries of topologically associated domains in schizophrenia. **2020**, 11, 1842	22
842	Comprehensive fundamental somatic variant calling and quality management strategies for human cancer genomes. **2021**, 22,	5
841	Uncovering Clinically Relevant Gene Fusions with Integrated Genomic and Transcriptomic Profiling of Metastatic Cancers. **2021**, 27, 522-531	4
840	Complex Variant Discovery Using Discordant Cluster Normalization. **2021**, 28, 185-194	

839	Detection of non-targeted transgenes by whole-genome resequencing for gene-doping control. **2021**, 28, 199-205	4
838	Detection of somatic structural variants from short-read next-generation sequencing data. **2021**, 22,	11
837	Epigenomic, genomic, and transcriptomic landscape of schwannomatosis. **2021**, 141, 101-116	11
836	H3 G34-mutant high-grade glioma. **2021**, 38, 4-13	7
835	Adaptation, ancestral variation and gene flow in a 'Sky Island' Drosophila species. **2021**, 30, 83-99	2
834	The Evolutionary Dynamics of Genetic Incompatibilities Introduced by Duplicated Genes in Arabidopsis thaliana. **2021**, 38, 1225-1240	0
833	Enhancing acetic acid and 5-hydroxymethyl furfural tolerance of C. saccharoperbutylacetonicum through adaptive laboratory evolution. **2021**, 101, 179-189	4
832	Long-read trio sequencing of individuals with unsolved intellectual disability. **2021**, 29, 637-648	7
831	A Performance Comparison of Commonly Used Assays to Detect RET Fusions. **2021**, 27, 1316-1328	11
830	Taking the next-gen step: Comprehensive antimicrobial resistance detection from Burkholderia pseudomallei. **2021**, 63, 103152	5
829	An Integrative DNA Sequencing and Methylation Panel to Assess Mismatch Repair Deficiency. **2021**, 23, 242-252	5
828	A New Catalog of Structural Variants in 1,301 A.□thaliana Lines from Africa, Eurasia, and North America Reveals a Signature of Balancing Selection at Defense Response Genes. **2021**, 38, 1498-1511	8
827	A paternally inherited 1.4 kb deletion of the 11p15.5 imprinting center 2 is associated with a mild familial Silver-Russell syndrome phenotype. **2021**, 29, 447-454	1
826	Overexpression of schizophrenia susceptibility factor human complement C4A promotes excessive synaptic loss and behavioral changes in mice. **2021**, 24, 214-224	49
825	Few Fixed Variants between Trophic Specialist Pupfish Species Reveal Candidate Cis-Regulatory Alleles Underlying Rapid Craniofacial Divergence. **2021**, 38, 405-423	4
824	BL1391: an established cell line from a human malignant peripheral nerve sheath tumor with unique genomic features. **2021**, 34, 238-245	0
823	Deletion Detection Method Using the Distribution of Insert Size and a Precise Alignment Strategy. **2021**, 18, 1070-1081	
822	Network-based analysis of allele frequency distribution among multiple populations identifies adaptive genomic structural variants.	0

821	Transposable elements contribute to genome dynamics and gene expression variation in the fungal plant pathogen Verticillium dahliae.	0
820	Detecting Causal Variants in Mendelian Disorders Using Whole-Genome Sequencing. **2021**, 2243, 1-25	2
819	Nebula: ultra-efficient mapping-free structural variant genotyper. **2021**, 49, e47	6
818	In vivo cytidine base editing of hepatocytes without detectable off-target mutations in RNA and DNA. **2021**, 5, 179-189	20
817	State-of-the-art structural variant calling: What went conceptually wrong and how to fix it?.	0
816	The genomes of precision edited cloned calves show no evidence for off-target events or increased de novo mutagenesis.	
815	Pathogenic Germline Variants in Cancer Susceptibility Genes in Children and Young Adults With Rhabdomyosarcoma. **2021**, 5,	6
814	Conservation and Loss of a Putative Iron Utilization Gene Cluster among Genotypes of. **2021**, 9,	
813	Next Generation Sequencing analysis suggests varied multistep mutational pathogenesis for Endocrine Mucin Producing Sweat Gland Carcinoma with comments on INSM1 and MUC2 suggesting a conjunctival origin. **2021**,	2
812	Ultraviolet radiation drives mutations in a subset of mucosal melanomas. **2021**, 12, 259	10
811	Genome structure variation analyses of peach reveal population dynamics and a 1.67 Mb causal inversion for fruit shape. **2021**, 22, 13	16
810	Methods to Study Genomic DNA Sequence Variation. **2021**, 59-92	
809	The antimalarial efficacy and mechanism of resistance of the novel chemotype DDD01034957. **2021**, 11, 1888	0
808	Whole genome sequencing of skull-base chordoma reveals genomic alterations associated with recurrence and chordoma-specific survival. **2021**, 12, 757	14
807	Rapid divergence of the male reproductive proteins in the Drosophila dunni group and implications for postmating incompatibilities between species. **2021**, 11,	0
806	Discovering single nucleotide variants and indels from bulk and single-cell ATAC-seq.	
805	Size Variation of the Nonrecombining Region on the Mating-Type Chromosomes in the Fungal Podospora anserina Species Complex. **2021**, 38, 2475-2492	0
804	ClinSV: clinical grade structural and copy number variant detection from whole genome sequencing data. **2021**, 13, 32	9

803	Analysis of Structural Variants Reveal Novel Selective Regions in the Genome of Meishan Pigs by Whole Genome Sequencing. **2021**, 12, 550676	4
802	Clinical and Biological Subtypes of B-cell Lymphoma Revealed by Microenvironmental Signatures. **2021**, 11, 1468-1489	27
801	A variant selection framework for genome graphs.	0
800	Clinically significant genomic alterations in the Chinese and Western patients with intrahepatic cholangiocarcinoma. **2021**, 21, 152	3
799	Whole genome sequencing of 45 Japanese patients with intellectual disability. **2021**, 185, 1468-1480	6
798	PopDel identifies medium-size deletions simultaneously in tens of thousands of genomes. **2021**, 12, 730	4
797	Genome sequences of Tropheus moorii and Petrochromis trewavasae, two eco-morphologically divergent cichlid fishes endemic to Lake Tanganyika. **2021**, 11, 4309	2
796	The lncRNA 44s2 Study Applicability to the Design of 45-55 Exon Skipping Therapeutic Strategy for DMD. **2021**, 9,	1
795	Intronic variant in POU1F1 associated with canine pituitary dwarfism. **2021**, 140, 1553-1562	0
794	Ancient Migrations - The first complete genome assembly, annotation and variants of the Zoroastrian-Parsi community of India.	
793	The impact of genomic structural variation on the transcriptome, chromatin, and proteome in the human brain.	1
792	A missense variant in IFT122 associated with a canine model of retinitis pigmentosa. **2021**, 140, 1569-1579	0
791	Association Between Mutations and Glucocorticoid Resistance in Children With Acute Lymphoblastic Leukemia. **2021**, 12, 634956	1
790	Genomic analyses provide insights into peach local adaptation and responses to climate change. **2021**, 31, 592-606	9
789	Transformed Canine and Murine Mesenchymal Stem Cells as a Model for Sarcoma with Complex Genomics. **2021**, 13,	3
788	Reconstructing the Lineage Histories and Differentiation Trajectories of Individual Cancer Cells in Myeloproliferative Neoplasms. **2021**, 28, 514-523.e9	42
787	An international virtual hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. **2021**, 10, 246	2
786	HolistIC: Leveraging Hi-C and Whole Genome Shotgun Sequencing for Double Minute Chromosome Discovery.	1

785	Leukemic stem cell phenotype is associated with mutational profile in acute myeloid leukemia. **2021**, 36, 401-412	3
784	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. **2021**, 23, 358-371	1
783	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology. **2021**, 5, 15	3
782	Genome-wide investigation identifies a rare copy-number variant burden associated with human spina bifida. **2021**, 23, 1211-1218	3
781	Shotgun transcriptome, spatial omics, and isothermal profiling of SARS-CoV-2 infection reveals unique host responses, viral diversification, and drug interactions. **2021**, 12, 1660	60
780	Molecular organization of recombinant human-Arabidopsis chromosomes in hybrid cell lines. **2021**, 11, 7160	0
779	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. **2021**, 11,	2
778	JAX-CNV: A whole genome sequencing-based algorithm for copy number detection at clinical grade level.	1
777	Mako: a graph-based pattern growth approach to detect complex structural variants.	
776	Comparison of Structural and Short Variants Detected by Linked-Read and Whole-Exome Sequencing in Multiple Myeloma. **2021**, 13,	1
775	Finding underlying genetic mechanisms of two patients with autism spectrum disorder carrying familial apparently balanced chromosomal translocations. **2021**, 23, e3322	
774	Computational Methods for Detecting Large-Scale Structural Rearrangements in Chromosomes. 37-51	
773	Combinatorial patterns of gene expression changes contribute to variable expressivity of the developmental delay-associated 16p12.1 deletion.	
772	A loss-of-function mutation in RORB disrupts saltatorial locomotion in rabbits. **2021**, 17, e1009429	1
771	Structural variant selection for high-altitude adaptation using single-molecule long-read sequencing.	1
770	Molecular characterization of DICER1-mutated pituitary blastoma. **2021**, 141, 929-944	6
769	The transcriptional landscape of Shh medulloblastoma. **2021**, 12, 1749	7
768	SENSV: Detecting Structural Variations with Precise Breakpoints using Low-Depth WGS Data from a Single Oxford Nanopore MinION Flowcell.	1

767	The Usefulness of Cell-Based and Liquid-Based Urine Tests in Clarifying the Diagnosis and Monitoring the Course of Urothelial Carcinoma. Identification of Novel, Potentially Actionable, and Somatic Mutations. **2021**, 11,	
766	A Population-Specific Major Allele Reference Genome From The United Arab Emirates Population. **2021**, 12, 660428	3
765	Integrated mutational landscape analysis of uterine leiomyosarcomas. **2021**, 118,	11
764	Proteogenomic and metabolomic characterization of human glioblastoma. **2021**, 39, 509-528.e20	71
763	Next Generation Sequencing Technology in the Clinic and Its Challenges. **2021**, 13,	7
762	Clinical and Genomic Characteristics of Adult Diffuse Midline Glioma. **2021**, 53, 389-398	2
761	Viola: a structural variant signature extractor with user-defined classifications.	
760	Cotton pan-genome retrieves the lost sequences and genes during domestication and selection. **2021**, 22, 119	10
759	Whole-genome resequencing of 445 Lactuca accessions reveals the domestication history of cultivated lettuce. **2021**, 53, 752-760	9
758	A comparison of tools for copy-number variation detection in germline whole exome and whole genome sequencing data.	2
757	Inversions maintain differences between migratory phenotypes of a songbird.	0
756	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. **2021**, 372,	100
755	A verified genomic reference sample for assessing performance of cancer panels detecting small variants of low allele frequency. **2021**, 22, 111	8
754	Integrative reconstruction of cancer genome karyotypes using InfoGenomeR. **2021**, 12, 2467	0
753	Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. **2021**, 4, 178-190	2
752	Protection of the C. elegans germ cell genome depends on diverse DNA repair pathways during normal proliferation. **2021**, 16, e0250291	3
751	Structural variants contribute to pangenome evolution of a plant pathogenic fungus.	
750	Extensive genome-wide duplications in the eastern oyster (). **2021**, 376, 20200164	5

749	In-frame deletion in canine PITRM1 is associated with a severe early-onset epilepsy, mitochondrial dysfunction and neurodegeneration. **2021**, 140, 1593-1609		2
748	Enabling Genomics Pipelines in Commodity Personal Computers With Flash Storage. **2021**, 12, 615958		
747	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. **2021**, 2, 100023-100023		7
746	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. **2021**, 108, 597-607		13
745	Reference SVA insertion polymorphisms are associated with Parkinson's Disease progression and differential gene expression. **2021**, 7, 44		5
744	Comparison of structural variants detected by optical mapping with long-read next-generation sequencing. *Bioinformatics*, **2021**,	7.2	1
743	Missense variant in LOXHD1 is associated with canine nonsyndromic hearing loss. **2021**, 140, 1611-1618		0
742	Detection of copy number variants in African goats using whole genome sequence data. **2021**, 22, 398		0
741	Simplifying the development of portable, scalable, and reproducible workflows.		
740	Familial Occurrence of Adult Granulosa Cell Tumors: Analysis of Whole-Genome Germline Variants. **2021**, 13,		1
739	Microdeletion of 9q22.3: A patient with minimal deletion size associated with a severe phenotype. **2021**, 185, 2070-2083		1
738	Chromosome-Level Assembly of the Atlantic Silverside Genome Reveals Extreme Levels of Sequence Diversity and Structural Genetic Variation. **2021**, 13,		2
737	Loose ends in cancer genome structure.		0
736	Dysgu: efficient structural variant calling using short or long reads.		1
735	The Telomere Length Landscape of Prostate Cancer.		
734	Different types of disease-causing noncoding variants revealed by genomic and gene expression analyses in families with X-linked intellectual disability. **2021**, 42, 835-847		
733	bric □brac controls sex pheromone choice by male European corn borer moths. **2021**, 12, 2818		5
732	Metastasis is altered through multiple processes regulated by the E2F1 transcription factor. **2021**, 11, 9502		2

731	Somatic structural variant formation is guided by and influences genome architecture.	1
730	Investigation of product-derived lymphoma following infusion of piggyBac-modified CD19 chimeric antigen receptor T cells. **2021**, 138, 1391-1405	26
729	Genomic insights into the pathogenesis of Epstein-Barr virus-associated diffuse large B-cell lymphoma by whole-genome and targeted amplicon sequencing. **2021**, 11, 102	6
728	Genomic Features and Classification of Homologous Recombination Deficient Pancreatic Ductal Adenocarcinoma. **2021**, 160, 2119-2132.e9	30
727	Genomic diversity in a population of Spodoptera frugiperda nucleopolyhedrovirus. **2021**, 90, 104749	5
726	Diverse tumorigenic consequences of human papillomavirus integration in primary oropharyngeal cancers.	
725	Control of gene doping in human and horse sports. **2021**,	5
724	GIP: An open-source computational pipeline for mapping genomic instability from protists to cancer cells.	
723	Transposable Elements Contribute to Genome Dynamics and Gene Expression Variation in the Fungal Plant Pathogen Verticillium dahliae. **2021**, 13,	2
722	2-kupl: mapping-free variant detection from DNA-seq data of matched samples. **2021**, 22, 304	
721	Alterations and Response to Immunotherapy in Solid Tumors. **2021**, 27, 4025-4035	6
720	Towards a Comprehensive Variation Benchmark for Challenging Medically-Relevant Autosomal Genes.	8
719	The genomes of precision edited cloned calves show no evidence for off-target events or increased de novo mutagenesis. **2021**, 22, 457	1
718	An integrated approach for copy number variation discovery in parent-offspring trios. **2021**, 22,	
717	Bi-allelic loss of ERGIC1 causes relatively mild arthrogryposis. **2021**, 100, 329-333	1
716	Evolution and genomic signatures of spontaneous somatic mutation in intestinal stem cells. **2021**, 31, 1419-1432	3
715	A cattle graph genome incorporating global breed diversity.	2
714	Comprehensive identification of transposable element insertions using multiple sequencing technologies. **2021**, 12, 3836	7

713	Genomic diversity, chromosomal rearrangements, and interspecies hybridization in the Ogataea polymorpha species complex. **2021**, 11,		0
712	Enhancer Hijacking Drives Oncogenic Expression in Lineage-Ambiguous Stem Cell Leukemia. **2021**, 11, 2846-2867		12
711	Extrachromosomal DNA is associated with chromothripsis events and diverse prognoses in gastric cardia adenocarcinoma.		1
710	A variant selection framework for genome graphs. *Bioinformatics*, **2021**, 37, i460-i467	7.2	1
709	Identification and characterization of inheritable structural variations induced by ion beam radiations in rice. **2021**, 823, 111757		0
708	Optical genome mapping identifies a germline retrotransposon insertion in SMARCB1 in two siblings with atypical teratoid rhabdoid tumors. **2021**, 255, 202-211		4
707	Lineage-defined leiomyosarcoma subtypes emerge years before diagnosis and determine patient survival. **2021**, 12, 4496		4
706	Recent ultra-rare inherited variants implicate new autism candidate risk genes. **2021**, 53, 1125-1134		13
705	Discovering single nucleotide variants and indels from bulk and single-cell ATAC-seq. **2021**, 49, 7986-7994		2
704	How Important Are Structural Variants for Speciation?. **2021**, 12,		3
703	NPSV: A simulation-driven approach to genotyping structural variants in whole-genome sequencing data. **2021**, 10,		2
702	Mako: A Graph-based Pattern Growth Approach to Detect Complex Structural Variants. **2021**,		2
701	Antibiotic Cycling Reverts Extensive Drug Resistance in Burkholderia multivorans. **2021**, 65, e0061121		2
700	Variant calling tool evaluation for variable size indel calling from next generation whole genome and targeted sequencing data.		1
699	Current status of structural variation studies in plants. **2021**, 19, 2153-2163		7
698	Endogenous protein tagging in medaka using a simplified CRISPR/Cas9 knock-in approach.		1
697	Retinoblastoma from human stem cell-derived retinal organoids. **2021**, 12, 4535		5
696	Genome-Wide Detection of Structural Variations Reveals New Regions Associated with Domestication in Small Ruminants. **2021**, 13,		0

695	In vivo modelling of patient genetic heterogeneity identifies concurrent Wnt and PI3K activity as a potent driver of invasive cholangiocarcinoma growth.	
694	GRIDSS2: comprehensive characterisation of somatic structural variation using single breakend variants and structural variant phasing. **2021**, 22, 202	5
693	Neuroblastoma Risk Assessment and Treatment Stratification with Hybrid Capture-Based Panel Sequencing. **2021**, 11,	
692	Model-guided development of an evolutionarily stable yeast chassis. **2021**, 17, e10253	3
691	Global mapping of cancers: The Cancer Genome Atlas and beyond. **2021**, 15, 2823-2840	10
690	WNT2 activation through proximal germline deletion predisposes to small intestinal neuroendocrine tumors and intestinal adenocarcinomas. **2021**, 30, 2429-2440	1
689	GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing.	1
688	R-loops and regulatory changes in chronologically ageing fission yeast cells drive non-random patterns of genome rearrangements. **2021**, 17, e1009784	
687	Specific BRCA and immune configurations determine optimal response to platinum-based chemotherapy in triple negative breast and ovarian carcinomas.	
686	A Comprehensive Map of Mycobacterium tuberculosis Complex Regions of Difference. **2021**, 6, e0053521	2
685	PanSVR: Pan-Genome Augmented Short Read Realignment for Sensitive Detection of Structural Variations. **2021**, 12, 731515	0
684	Severe inbreeding and gene loss in the historical and extant population of the critically endangered Devils Hole pupfish.	1
683	Methods and Developments in Graphical Pangenomics. **2021**, 101, 1-14	0
682	Discovery of structural deletions in breast cancer predisposition genes using whole genome sequencing data from > 2000 women of African-ancestry. **2021**, 140, 1449-1457	1
681	From DNA human sequence to the chromatin higher order organisation and its biological meaning: Using biomolecular interaction networks to understand the influence of structural variation on spatial genome organisation and its functional effect. **2021**, 121, 171-171	0
680	Metagenomic analysis of primary colorectal carcinomas and their metastases identifies potential microbial risk factors. **2021**, 15, 3363-3384	4
679	New high-quality peach (Prunus persica L. Batsch) genome assembly to analyze the molecular evolutionary mechanism of volatile compounds in peach fruits. **2021**, 108, 281-295	3
678	Aperture: alignment-free detection of structural variations and viral integrations in circulating tumor DNA. **2021**, 22,	1

677	A Homolog of the Gene Is□nvolved in Nonhost Resistance to Wheat Stem Rust in. **2021**, MPMI06210137R	0
676	Identification of candidate genes associated with slow-melting flesh trait in peach using bulked segregant analysis and RNA-seq. **2021**, 286, 110208	3
675	Clonal dynamics in early human embryogenesis inferred from somatic mutation. **2021**, 597, 393-397	17
674	Canine oral melanoma genomic and transcriptomic study defines two molecular subgroups with different therapeutical targets.	
673	Copy number signatures predict chromothripsis and clinical outcomes in newly diagnosed multiple myeloma. **2021**, 12, 5172	2
672	Data-driven approaches to advance research and clinical care for pediatric cancer. **2021**, 1876, 188571	1
671	Simulated validation of intron-less transgene detection using DELLY for gene-doping control in horse sports. **2021**, 52, 759-761	0
670	AR gene rearrangement analysis in liquid biopsies reveals heterogeneity in lethal prostate cancer. **2021**, 28, 645-655	0
669	Genome sequencing data analysis for rare disease gene discovery. **2021**,	2
668	Genomic characterization and outcome evaluation of kinome fusions in lung cancer revealed novel druggable fusions. **2021**, 5, 81	2
667	Genome sequencing as a first-line diagnostic test for hospitalized newborns.	
666	Deciphering complex genome rearrangements in C. elegans using short-read whole genome sequencing. **2021**, 11, 18258	2
665	Molecular characterisation of pancreatic ductal adenocarcinoma with fusions and review of the literature. **2021**,	2
664	An ensemble deep learning framework to refine large deletions in linked-reads.	1
663	CloneSig can jointly infer intra-tumor heterogeneity and mutational signature activity in bulk tumor sequencing data. **2021**, 12, 5352	1
662	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. **2021**, 108, 1981-2005	4
661	Systematic discovery of gene fusions in pediatric cancer by integrating RNA-seq and WGS.	2
660	Expanding the conservation genomics toolbox: Incorporating structural variants to enhance genomic studies for species of conservation concern. **2021**, 30, 5949-5965	3

659	Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. **2021**, 39, 1129-1140		10
658	Generation of iPSC lines with high cytogenetic stability from peripheral blood mononuclear cells (PBMCs).		
657	Disentangling heterogeneity of Malignant Pleural Mesothelioma through deep integrative omics analyses.		0
656	Multiplatform discovery and regulatory function analysis of structural variations in non-small cell lung carcinoma. **2021**, 36, 109660		0
655	Multiplexed Simian Immunodeficiency Virus-Specific Paired RNA-Guided Cas9 Nickases Inactivate Proviral DNA. **2021**, 95, e0088221		0
654	Genome Structural Variation Landscape and Its Selection Signatures in the Fast-growing Strains of the Pacific Oyster, Crassostrea gigas. **2021**, 23, 736-748		1
653	CNproScan: Hybrid CNV detection for bacterial genomes. **2021**, 113, 3103-3111		
652	SimFFPE and FilterFFPE: improving structural variant calling in FFPE samples. **2021**, 10,		3
651	Viola: a structural variant signature extractor with user-defined classifications. *Bioinformatics*, **2021**,	7.2	0
650	Comprehensive analysis of mutational signatures in pediatric cancers.		0
649	Genomic landscape and evolution of arm aneuploidy in lung adenocarcinoma. **2021**, 23, 870-878		0
648	An international virtual hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. **2021**, 10, 246		1
647	TT-Mars: Structural Variants Assessment Based on Haplotype-resolved Assemblies.		1
646	Large-Scale Screening for Monogenic and Clinically Defined Familial Hypercholesterolemia in Iceland. **2021**, 41, 2616-2628		4
645	Human Genetics of Face Recognition: Discovery of MCTP2 Mutations in Humans with Face Blindness (Congenital Prosopagnosia).		
644	Cnngeno: A high-precision deep learning based strategy for the calling of structural variation genotype. **2021**, 94, 107417		0
643	The genome of the zoonotic malaria parasite Plasmodium simium reveals adaptations to host switching. **2021**, 19, 219		6
642	Related Inference: A Supervised Learning Approach to Detect Signal Variation in Genome Data. **2021**,		

641	Computational cancer genomics. **2021**, 329-359		
640	UV-exposure, endogenous DNA damage, and DNA replication errors shape the spectra of genome changes in human skin. **2021**, 17, e1009302		10
639	Bioinformatics workflows for clinical applications in precision oncology. **2021**,		1
638	Calling large indels in 1047 Arabidopsis with IndelEnsembler. **2021**, 49, 10879-10894		1
637	Population Genomics of Fungal Plant Pathogens and the Analyses of Rapidly Evolving Genome Compartments. **2020**, 2090, 337-355		4
636	Bioinformatics for Cancer Immunotherapy. **2020**, 2120, 1-9		6
635	Detecting Large Indels Using Optical Map Data. **2018**, 108-127		1
634	The SeqAn C++ template library for efficient sequence analysis: A resource for programmers. **2017**, 261, 157-168		49
633	Untangling a complex web: Computational analyses of tumor molecular profiles to decode driver mechanisms. **2020**, 47, 595-609		0
632	Butler enables rapid cloud-based analysis of thousands of human genomes. **2020**, 38, 288-292		9
631	A deep learning approach for filtering structural variants in short read sequencing data. **2021**, 22,		9
630	Bivartect: accurate and memory-saving breakpoint detection by direct read comparison. *Bioinformatics*, **2020**, 36, 2725-2730	7.2	2
629	Parliament2: Accurate structural variant calling at scale. **2020**, 9,		17
628	Corded and Hyalinized Endometrioid Adenocarcinoma (CHEC) of the Uterine Corpus are Characterized by CTNNB1 Mutations and Can Show Adverse Clinical Outcomes. **2021**, 40, 103-115		1
627	Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast.		1
626	SplitThreader: Exploration and analysis of rearrangements in cancer genomes.		8
625	SvABA: Genome-wide detection of structural variants and indels by local assembly.		5
624	GRIDSS: sensitive and specific genomic rearrangement detection using positional de Bruijn graph assembly.		4

623	The Sequence of 1504 Mutants in the Model Rice Variety Kitaake Facilitates Rapid Functional Genomic Studies.	1
622	deStruct: Accurate Rearrangement Detection using Breakpoint Specific Realignment.	4
621	HadoopCNV: A dynamic programming imputation algorithm to detect copy number variants from sequencing data.	1
620	Limited contribution of rare, noncoding variation to autism spectrum disorder from sequencing of 2,076 genomes in quartet families.	6
619	Mapping and phasing of structural variation in patient genomes using nanopore sequencing.	6
618	Large-Scale Uniform Analysis of Cancer Whole Genomes in Multiple Computing Environments.	14
617	Accurate detection of complex structural variations using single molecule sequencing.	33
616	Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line.	6
615	npInv: accurate detection and genotyping of inversions mediated by non-allelic homologous recombination using long read sub-alignment.	1
614	Enabling rapid cloud-based analysis of thousands of human genomes via Butler.	2
613	Identifying structural variants using linked-read sequencing data.	5
612	Multi-platform discovery of haplotype-resolved structural variation in human genomes.	26
611	Fast and Accurate Genomic Analyses using Genome Graphs.	8
610	Somatic inactivating PTPRJ mutations and dysregulated pathways identified in canine melanoma by integrated comparative genomic analysis.	1
609	Detection of complex structural variation from paired-end sequencing data.	5
608	Revealing the impact of recurrent and rare structural variants in multiple myeloma.	1
607	Bovine breed-specific augmented reference graphs facilitate accurate sequence read mapping and unbiased variant discovery.	2
606	A new domestic cat genome assembly based on long sequence reads empowers feline genomic medicine and identifies a novel gene for dwarfism.	12

605	Genomic analyses provide insights into peach local adaptation and responses to climate change.	1
604	A comprehensive benchmarking of WGS-based structural variant callers.	3
603	Shotgun Transcriptome and Isothermal Profiling of SARS-CoV-2 Infection Reveals Unique Host Responses, Viral Diversification, and Drug Interactions. **2020**,	51
602	Reference genome for the highly transformable Setaria viridis cultivar ME034V.	0
601	Highly accurate long-read HiFi sequencing data for five complex genomes.	2
600	Rapid divergence of the copulation proteins in theDrosophila dunnigroup is associated with hybrid post-mating-prezygotic incompatibilities.	3
599	Long-read genome sequencing for the diagnosis of neurodevelopmental disorders.	4
598	GRIDSS2: comprehensive characterisation of somatic structural variation using single breakend variants and structural variant phasing.	6
597	Evolution and genomic signatures of spontaneous somatic mutation in Drosophila intestinal stem cells.	1
596	Multi-Platform Assessment of DNA Sequencing Performance using Human and Bacterial Reference Genomes in the ABRF Next-Generation Sequencing Study.	1
595	The evolutionary dynamics of genetic incompatibilities introduced by duplicated genes in Arabidopsis thaliana.	1
594	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families.	1
593	Ultra-fast Prediction of Somatic Structural Variations by Reduced Read Mapping via Pan-Genome k-mer Sets.	0
592	Chromosome-level assembly of the Atlantic silverside genome reveals extreme levels of sequence diversity and structural genetic variation.	1
591	Pangenome-based genome inference.	11
590	Clonal dynamics in early human embryogenesis inferred from somatic mutation.	0
589	Whole Genomes Define Concordance of Matched Primary, Xenograft, and Organoid Models of Pancreas Cancer.	2
588	ACEseq allele specific copy number estimation from whole genome sequencing.	17

587	Deficiency of nucleotide excision repair explains mutational signature observed in cancer.	2
586	Whole genome and transcriptome maps of the entirely black native Korean chicken breed Yeonsan Ogye.	1
585	The Tandem Duplicator Phenotype is a prevalent genome-wide cancer configuration driven by distinct gene mutations.	1
584	Whole-genome resequencing and pan-transcriptome reconstruction highlight the impact of genomic structural variation on secondary metabolism gene clusters in the grapevine Esca pathogenPhaeoacremonium minimum.	1
583	A large-scale whole-genome sequencing analysis reveals highly specific genome editing by both Cas9 and Cpf1 nucleases in rice.	2
582	Integrated sequence and gene expression analysis of mouse models of breast cancer reveals critical events with human parallels.	2
581	Characterization of segmental duplications and large inversions using Linked-Reads.	4
580	LinkedSV for detection of mosaic structural variants from linked-read exome and genome sequencing data.	1
579	Parliament2: Fast Structural Variant Calling Using Optimized Combinations of Callers.	12
578	GenPipes: an open-source framework for distributed and scalable genomic analyses.	2
577	duphold: scalalable, depth-based annotation and curation of high-confidence structural variant calls.	0
576	Structural variants, clonal propagation, and genome evolution in grapevine (Vitis vinifera).	11
575	Highly-accurate long-read sequencing improves variant detection and assembly of a human genome.	29
574	MoMI-G: Modular Multi-scale Integrated Genome Graph Browser.	1
573	Kevlar: a mapping-free framework for accurate discovery ofde novovariants.	1
572	Reconstruction of clone- and haplotype-specific cancer genome karyotypes from bulk tumor samples.	5
571	Integrated Phosphoproteomics and Transcriptional Classifiers Reveal Hidden RAS Signaling Dynamics in Multiple Myeloma.	1
570	The Tangent copy-number inference pipeline for cancer genome analyses.	3

569	Nebula: Ultra-efficient mapping-free structural variant genotyper.	3
568	The genomic diversification of clonally propagated grapevines.	6
567	Identification of Pathogenic Structural Variants in Rare Disease Patients through Genome Sequencing.	8
566	Paragraph: A graph-based structural variant genotyper for short-read sequence data.	8
565	Genotyping structural variants in pangenome graphs using the vg toolkit.	12
564	HiNT: a computational method for detecting copy number variations and translocations from Hi-C data.	2
563	NanoVar: Accurate Characterization of Patients□Genomic Structural Variants Using Low-Depth Nanopore Sequencing.	2
562	An integrated personal and population-based Egyptian genome reference.	0
561	Functional annotation of rare structural variation in the human brain.	3
560	Enhancing sensitivity and controlling false discovery rate in somatic indel discovery.	1
559	MapCaller □An integrated and efficient tool for short-read mapping and variant calling using high-throughput sequenced data.	4
558	CloneSig can jointly infer intra-tumor heterogeneity and mutational signature activity in bulk tumor sequencing data.	2
557	The genome of the zoonotic malaria parasite Plasmodium simium reveals adaptations to host-switching.	5
556	Diverse noncoding mutations contribute to deregulation of cis-regulatory landscape in pediatric cancers.	1
555	Genotyping structural variants in pangenome graphs using the vg toolkit. **2020**, 21, 35	65
554	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. **2017**, 6, 664	37
553	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. **2017**, 6, 664	33
552	Breaking point: the genesis and impact of structural variation in tumours. **2018**, 7,	5

551	Integrative analysis of structural variations using short-reads and linked-reads yields highly specific and sensitive predictions. **2020**, 16, e1008397	2
550	Genome-Wide Analysis Identifies Germ-Line Risk Factors Associated with Canine Mammary Tumours. **2016**, 12, e1006029	13
549	The Impact of Environmental and Endogenous Damage on Somatic Mutation Load in Human Skin Fibroblasts. **2016**, 12, e1006385	55
548	Two extended haplotype blocks are associated with adaptation to high altitude habitats in East African honey bees. **2017**, 13, e1006792	42
547	PEA15 loss of function and defective cerebral development in the domestic cat. **2020**, 16, e1008671	2
546	A new domestic cat genome assembly based on long sequence reads empowers feline genomic medicine and identifies a novel gene for dwarfism. **2020**, 16, e1008926	29
545	SoftSearch: integration of multiple sequence features to identify breakpoints of structural variations. **2013**, 8, e83356	30
544	Whole genome sequencing based characterization of extensively drug-resistant Mycobacterium tuberculosis isolates from Pakistan. **2015**, 10, e0117771	41
543	XomAnnotate: Analysis of Heterogeneous and Complex Exome- A Step towards Translational Medicine. **2015**, 10, e0123569	5
542	Improving the Power of Structural Variation Detection by Augmenting the Reference. **2015**, 10, e0136771	6
541	Comparative Genome of GK and Wistar Rats Reveals Genetic Basis of Type 2 Diabetes. **2015**, 10, e0141859	11
540	Identifying Likely Transmission Pathways within a 10-Year Community Outbreak of Tuberculosis by High-Depth Whole Genome Sequencing. **2016**, 11, e0150550	15
539	AMYCNE: Confident copy number assessment using whole genome sequencing data. **2018**, 13, e0189710	11
538	Insertion and deletion evolution reflects antibiotics selection pressure in a Mycobacterium tuberculosis outbreak. **2020**, 16, e1008357	5
537	Characterization of Structural Variations in the Context of 3D Chromatin Structure. **2019**, 42, 512-522	9
536	The root hair defective phenotype of Pol IV subunit mutant is associated with a deletion in RHD6. **2019**, 2019,	1
535	Next generation mapping reveals novel large genomic rearrangements in prostate cancer. **2017**, 8, 23588-23602	30
534	Frequent L1 retrotranspositions originating from TTC28 in colorectal cancer. **2014**, 5, 853-9	40

533	Detection of circulating tumor DNA in patients with osteosarcoma. **2018**, 9, 12695-12704	26
532	Unscrambling the genomic chaos of osteosarcoma reveals extensive transcript fusion, recurrent rearrangements and frequent novel TP53 aberrations. **2016**, 7, 5273-88	45
531	Immortalization capacity of HPV types is inversely related to chromosomal instability. **2016**, 7, 37608-37621	17
530	A framework for generating interactive reports for cancer genome analysis. **2017**, 2, 457	1
529	MVSC: A Multi-variation Simulator of Cancer Genome. **2020**, 23, 326-333	1
528	Defind: Detecting Genomic Deletions by Integrating Read Depth, GC Content, Mapping Quality and Paired-end Mapping Signatures of Next Generation Sequencing Data. **2019**, 14, 130-138	2
527	Genomic Landscape of Young-Onset Bladder Cancer and Its Prognostic Implications on Adult Bladder Cancer. **2020**, 12,	0
526	An improved understanding of cancer genomics through massively parallel sequencing. **2014**, 3, 243-259	8
525	Assembling and Validating Bioinformatic Pipelines for Next-Generation Sequencing Clinical Assays. **2020**, 144, 1118-1130	7
524	Diversification of the heat shock response by Helitron transposable elements. **2019**, 8,	8
523	Variant profiling of evolving prokaryotic populations. **2017**, 5, e2997	11
522	sv-callers: a highly portable parallel workflow for structural variant detection in whole-genome sequence data. **2020**, 8, e8214	7
521	GROM-RD: resolving genomic biases to improve read depth detection of copy number variants. **2015**, 3, e836	19
520	intansv: an R package for integrative analysis of structural variations. **2020**, 8, e8867	3
519	Enhancing breakpoint resolution with deep segmentation model: A general refinement method for read-depth based structural variant callers. **2021**, 17, e1009186	
518	Combinatorial patterns of gene expression changes contribute to variable expressivity of the developmental delay-associated 16p12.1 deletion. **2021**, 13, 163	0
517	Influences of rare copy number variation on human complex traits.	0
516	Simplifying the development of portable, scalable, and reproducible workflows. **2021**, 10,	0

515	Genetic alteration and clonal evolution of primary glioblastoma into secondary gliosarcoma. **2021**, 27, 1483-1492	3
514	C. elegans genome-wide analysis reveals DNA repair pathways that act cooperatively to preserve genome integrity upon ionizing radiation. **2021**, 16, e0258269	
513	Targeting the Non-Coding Genome for the Diagnosis of Disorders of Sex Development. **2021**, 1-19	1
512	A Benchmark of Structural Variant Analysis Tools for Next Generation Sequencing Data. **2013**, 2, 86-98	
511	SVEM: A Structural Variant Estimation Method Using Multi-mapped Reads on Breakpoints. **2014**, 208-219	
510	Bioinformatics Analysis of Sequence Data. **2016**, 317-333	
509	Detecting Horizontal Gene Transfer by Mapping Sequencing Reads Across Species Boundaries.	
508	Allele-Specific Quantification of Structural Variations in Cancer Genomes.	1
507	Genotyping of Inversions and Tandem Duplications.	
506	HySA: A Hybrid Structural variant Assembly approach using next generation and single-molecule sequencing technologies.	1
505	Genomic Rearrangements Considered as Quantitative Traits.	
504	Enhancing Knowledge Discovery from Cancer Genomics Data with Galaxy.	
503	A computational method for detection of structural variants using Deviant Reads and read pair Orientation: DevRO.	
502	A recurrence based approach for validating structural variation using long-read sequencing technology.	
501	Enhancing sensitivity and controlling false discovery rate in somatic indel discovery using a latent variable model.	
500	HiPiler: Visual Exploration of Large Genome Interaction Matrices with Interactive Small Multiples.	1
499	Informatics for Cancer Immunotherapy.	1
498	Computational proteogenomic identification and functional interpretation of translated fusions and micro structural variations in cancer.	

497 SQUID: Transcriptomic Structural Variation Detection from RNA-seq.

496 Generation, selection and transcriptomic profiling of human neuromesodermal and spinal cord progenitors in vitro. 2

495 A framework for generating interactive reports for cancer genome analysis.

494 Recurrent loss of heterozygosity correlates with clinical outcome in pancreatic neuroendocrine cancer.

493 Combining accurate tumour genome simulation with crowd-sourcing to benchmark somatic structural variant detection. 1

492 SVEngine: an efficient and versatile simulator of genome structural variations with features of cancer clonal evolution.

491 An Integrated Method of Detecting Copy Number Variation Based on Sequence Assembly. **2018**, 589-594

490 PASA: Identifying More Credible Structural Variants of Hedou12. **2018**, 553-558

489 Improved Indel Detection Algorithm Based on Split-Read and Read-Depth. **2018**, 853-864

488 Integration of Genomic and Transcriptomic Features in Pancreatic Cancer Reveals Increased Cell Cycle Progression in Metastases.

487 A Randomized Iterative Approach for SV Discovery with SVelter. **2018**, 1833, 169-177

486 SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. 1

485 Cluster expansion of apolipoprotein D (ApoD) genes in teleost fishes.

484 P. falciparum PfATP4 Multi-Drug Resistance Resistance to KAE609 (Cipargamin) is Present in Africa.

483 Multiple myeloma immunoglobulin translocations portend poor prognosis. 1

482 Crispr/Cas9 Targeted Capture of Mammalian Genomic Regions for Characterization by NGS.

481 Discovery of tandem and interspersed segmental duplications using high throughput sequencing. 1

480 Detecting Large Indels Using Optical Map Data.

479	Origin and recent expansion of an endogenous gammaretroviral lineage in canids.	
478	Reconstructing and characterizing focal amplifications in cancer using AmpliconArchitect.	0
477	Retrotransposon insertions can initiate colorectal cancer and are associated with poor survival.	0
476	SVXplorer: Identification of structural variants through overlap of discordant clusters.	
475	Evaluation of the performance of copy number variant prediction tools for the detection of deletions from whole genome sequencing data.	
474	Analysis of structural variants in four African Cichlids highlights an association with developmental and immune related genes.	1
473	ReALLEN: structural variation discovery in cancer genome by sensitive analysis of single-end reads.	
472	simuG: a general-purpose genome simulator.	
471	Removal of a subset of non-essential genes fully attenuates a highly virulentMycoplasmastrain.	0
470	Guidelines for Bioinformatics and the Statistical Analysis of Omic Data. **2019**, 45-75	
469	Enhancing breakpoint resolution with deep segmentation model: a general refinement method for read-depth based structural variant callers.	
468	R-loops and regulatory changes in chronologically ageing fission yeast cells drive non-random patterns of genome rearrangements.	
467	JuLI: accurate detection of DNA fusions in clinical sequencing for precision oncology.	
466	DeepSV: Accurate calling of genomic deletions from high-throughput sequencing data using deep convolutional neural network.	
465	De novo diploid genome assembly for genome-wide structural variant detection.	1
464	Evaluation of computational genotyping of Structural Variations for clinical diagnoses.	
463	Whole Genome Detection of Sequence and Structural Polymorphism in Six Diverse Horses.	
462	Detection of genome-wide structural variations in the Shanghai Holstein cattle population using next-generation sequencing. **2019**, 32, 320-333	0

461	Nbs1 Mediates Assembly and Activity of the Mre11 complex.	
460	BreakCA, a method to discover indels using ChIP-seq and ATAC-seq reads, finds recurrent indels in regulatory regions of neuroblastoma genomes.	
459	Low Abundance of Circulating Tumor DNA in Localized Prostate Cancer.	1
458	Insertional oncogenesis by HPV70 revealed by multiple genomic analyses in a clinically HPV-negative cervical cancer.	
457	Genomic basis of circannual rhythm in the european corn borer moth.	0
456	Shiny-SoSV: A web-based performance calculator for somatic structural variant detection.	
455	Mutational signatures are jointly shaped by DNA damage and repair.	3
454	Discovery and Quality Analysis of a Comprehensive Set of Structural Variants and Short Tandem Repeats.	2
453	Detecting Transcriptomic Structural Variants in Heterogeneous Contexts via the Multiple Compatible Arrangements Problem.	
452	Hecaton: reliably detecting copy number variation in plant genomes using short read sequencing data.	
451	Long-read assembly of the Chinese rhesus macaque genome and identification of ape-specific structural variants.	
450	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants.	1
449	PopDel identifies medium-size deletions jointly in tens of thousands of genomes.	0
448	Genome sequencing for early-onset dementia: high diagnostic yield and frequent observation of multiple contributory alleles.	1
447	Detection and characterization of copy number variants based on whole-genome sequencing by DNBSEQ platforms.	
446	Long-read-based Human Genomic Structural Variation Detection with cuteSV.	1
445	Genetic Basis ofDe NovoAppearance of Carotenoid Ornamentation in Bare-Parts of Canaries.	
444	Integrative genomic, transcriptomic, and epigenomic analyses of benign prostatic hyperplasia reveal new options for therapy.	

443	Detection of somatic structural variants from short-read next-generation sequencing data.	1
442	Single cell tri-channel-processing reveals structural variation landscapes and complex rearrangement processes.	1
441	The population genomics of structural variation in a songbird genus.	2
440	Noncoding de novo mutations contribute to autism spectrum disorder via chromatin interactions.	0
439	Analysis of unmapped regions associated with long deletions in Korean whole genome sequences based on short read data. **2019**, 17, e40	0
438	Metastasis is altered through multiple processes regulated by the E2F1 transcription factor.	
437	A 63-bp insertion in exon 2 of the porcine KIF21A gene is associated with arthrogryposis multiplex congenita.	0
436	Adaptation, ancestral variation and gene flow in a Sky IslandDrosophilaspecies.	2
435	Integrated analysis of whole genome and transcriptome sequencing in a young patient with gastric cancer provides insights for precision therapy. **2020**, 20, 115	
434	Massive parallel sequencing in individuals with multiple primary tumours reveals the benefit of re-analysis. **2021**, 19, 46	1
433	Similarity-based analysis of allele frequency distribution among multiple populations identifies adaptive genomic structural variants. **2021**,	1
432	A cis-regulatory element promoting increased transcription at low temperature in cultured ectothermic Drosophila cells. **2021**, 22, 771	0
431	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the Gene. **2021**, 7, e632	3
430	Insertion and deletion evolution reflects antibiotics selection pressure in a Mycobacterium tuberculosis outbreak.	1
429	Chasing breeding footprints through structural variations in Cucumis melo and wild relatives. **2021**, 11,	1
428	Aperture: Accurate detection of structural variations and viral integrations in circulating tumor DNA using an alignment-free algorithm.	0
427	Evolutionary genomics of structural variation in Asian rice (Oryza sativa) and its wild progenitor (O. rufipogon).	2
426	Activation of cryptic splicing in bovine WDR19 is associated with reduced semen quality and male fertility.	

425	Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication.	
424	Analysis of structural variants in four African Cichlids highlights an association with developmental and immune related genes.	
423	Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication.	
422	PEA15 loss of function and defective cerebral development in the domestic cat.	
421	Disruption of the topological associated domain at Xp21.2 is related to gonadal dysgenesis: A general mechanism of pathogenesis.	
420	Detecting genomic deletions from high-throughput sequence data with unsupervised learning.	
419	Conspicuous candidate alleles point to cis-regulatory divergence underlying rapidly evolving craniofacial phenotypes.	
418	CNV-PG: a machine-learning framework for accurate copy number variation predicting and genotyping.	0
417	Oncogenes hijack a constitutively activeTP53promoter in osteosarcoma.	1
416	BAdabouM: a genomic structural variations discovery tool for polymorphism analyses.	
415	A Genetic Model for Central Chondrosarcoma Evolution Correlates with Patient Outcome.	
414	Combining callers improves the detection of copy number variants from whole-genome sequencing. **2021**,	0
413	ClinSV: Clinical grade structural and copy number variant detection from whole genome sequencing data.	0
412	Sprites2: Detection of Deletions Based on an Accurate Alignment Strategy. **2018**, 139-150	
411	Acis-regulatory element promoting increased transcription at low temperature in cultured ectothermicDrosophilacells.	
410	Methods developed during the first National Center for Biotechnology Information Structural Variation Codeathon at Baylor College of Medicine. 9, 1141	
409	Genomic Diversity in a Population of Spodoptera frugiperda Nucleopolyhedrovirus.	
408	Population differentiation and structural variation in the Manduca sexta genome across the United States.	0

407	Copy number signatures predict chromothripsis and associate with poor clinical outcomes in patients with newly diagnosed multiple myeloma.	
406	Evaluation of copy number variants for genetic hearing loss: a review of current approaches and recent findings. **2021**, 1	2
405	Whole-genome analysis of Nigerian patients with breast cancer reveals ethnic-driven somatic evolution and distinct genomic subtypes. **2021**, 12, 6946	1
404	The telomere length landscape of prostate cancer. **2021**, 12, 6893	0
403	Molecular subtyping of ependymoma and prognostic impact of Ki-67. **2021**, 1	2
402	The benefit of diagnostic whole genome sequencing in schizophrenia and other psychotic disorders. **2021**,	1
401	Comprehensive characterization of copy number variation (CNV) called from array, long- and short-read data. **2021**, 22, 826	0
400	APOBEC3A drives deaminase domain-independent chromosomal instability to promote pancreatic cancer metastasis.. **2021**, 2, 1338-1356	4
399	Focal amplifications are associated with chromothripsis events and diverse prognoses in gastric cardia adenocarcinoma. **2021**, 12, 6489	3
398	Cancer drivers and clonal dynamics in acute lymphoblastic leukaemia subtypes. **2021**, 11, 177	2
397	Extensive allele mining discovers novel genetic diversity in the loci controlling frost tolerance in barley. **2021**, 1	4
396	Intraspecies Genomic Divergence of a Fig Wasp Species Is Due to Geographical Barrier and Adaptation. **2021**, 9,	0
395	Rearrangement-mediated cis-regulatory alterations in advanced patient tumors reveal interactions with therapy. **2021**, 37, 110023	0
394	Targeted profiling of human extrachromosomal DNA by CRISPR-CATCH.	1
393	PerSVade: Personalized Structural Variation detection in your species of interest.	0
392	Patient-derived organoids as a preclinical platform for precision medicine in colorectal cancer. **2021** ,	3
391	Genome sequencing as a first-line diagnostic test for hospitalized infants.. **2021**,	0
390	CNV-P: a machine-learning framework for predicting high confident copy number variations.. **2021**, 9, e12564	0

389	BreakNet: detecting deletions using long reads and a deep learning approach. **2021**, 22, 577	0
388	Detecting inversions in routine molecular diagnosis in MMR genes.. **2022**, 1	
387	Detecting novel genomic structural variants through negative binomial optimization. **2020**,	0
386	Novel sequencing technologies and bioinformatic tools for deciphering the non-coding genome. **2021**, 33, 133-145	0
385	Xq27.1 palindrome mediated interchromosomal insertion likely causes familial congenital bilateral laryngeal abductor paralysis (Plott syndrome).. **2022**,	2
384	SAM-TB: a whole genome sequencing data analysis website for detection of Mycobacterium tuberculosis drug resistance and transmission.. **2022**,	1
383	Identification of processed pseudogenes in the genome of Thoroughbred horses: Possibility of gene-doping detection considering the presence of pseudogenes.. **2022**,	1
382	Canine Oral Melanoma Genomic and Transcriptomic Study Defines Two Molecular Subgroups with Different Therapeutical Targets.. **2022**, 14,	
381	De novo mutations identified by whole-genome sequencing implicate chromatin modifications in obsessive-compulsive disorder.. **2022**, 8, eabi6180	0
380	Dysgu: efficient structural variant calling using short or long reads.. **2022**,	0
379	Genome assembly, structural variants, and genetic differentiation between Lake Whitefish young species pairs (Coregonus sp.) with long and short reads.	0
378	Computational methods and translational applications for targeted next-generation sequencing platforms.. **2022**,	1
377	Convergent recruitment of Amh as the sex determination gene in two lineages of stickleback fish.	
376	JAX-CNV: A Whole Genome Sequencing-based Algorithm for Copy Number Detection at Clinical Grade Level.. **2022**,	
375	SCHOOL: Software for Clinical Health in Oncology for Omics Laboratories.. **2022**, 13, 1	0
374	Local adaptation of life cycles in a butterfly is associated with variation in several circadian clock genes.. **2021**,	2
373	Profilin 1 deficiency drives mitotic defects and impairs genome stability.	
372	GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing.. **2022**,	0

371	Very small spontaneous structural mutation rate in green algae.	0
370	Curated variation benchmarks for challenging medically relevant autosomal genes.. **2022**,	12
369	A platform for oncogenomic reporting and interpretation.. **2022**, 13, 756	1
368	Transient inhibition of the JAK/STAT pathway prevents B-ALL development in genetically predisposed mice.. **2022**,	0
367	Graph-based pan-genome reveals structural and sequence variations related to agronomic traits and domestication in cucumber.. **2022**, 13, 682	6
366	Effects of Mutations on Leukemogenesis and Targeting of Children With Acute Lymphoblastic Leukemia.. **2022**, 10, 712484	0
365	Clinical response to nivolumab in an INI1-deficient pediatric chordoma correlates with immunogenic recognition of brachyury.. **2021**, 5, 103	2
364	Pan-Genomic Sequencing Reveals Actionable Deletions and Kataegis in Anaplastic Thyroid Carcinoma.. **2021**, 13,	2
363	Cryptic MYC insertions in Burkitt lymphoma: New data and a review of the literature.. **2022**, 17, e0263980	0
362	The genetic landscape of pancreatic head ductal adenocarcinoma in China and prognosis stratification.. **2022**, 22, 186	3
361	Placental methylome reveals a 22q13.33 brain regulatory gene locus associated with autism.. **2022**, 23, 46	1
360	Long-read sequencing of diagnosis and post-therapy medulloblastoma reveals complex rearrangement patterns and epigenetic signatures.	
359	An ancient haplotype containing antimicrobial peptide gene variants is associated with severe fungal skin disease in Persian cats.. **2022**, 18, e1010062	0
358	Progress in Methods for Copy Number Variation Profiling.. **2022**, 23,	1
357	Retrotransposon instability dominates the acquired mutation landscape of mouse induced pluripotent stem cells.	0
356	Tool evaluation for the detection of variably sized indels from next generation whole genome and targeted sequencing data.. **2022**, 18, e1009269	0
355	Enhancer retargeting of CDX2 and UBTF::ATXN7L3 define a subtype of high-risk B-progenitor acute lymphoblastic leukemia.. **2022**,	1
354	Comparison of structural variants in the whole genome sequences of two Medicago truncatula ecotypes: Jemalong A17 and R108.. **2022**, 22, 77	1

353	Population differentiation and structural variation in the Manduca sexta genome across the United States.. **2022**,	0
352	A population study of clinically actionable genetic variation affecting drug response from the Middle East.. **2022**, 7, 10	5
351	A cattle graph genome incorporating global breed diversity.. **2022**, 13, 910	3
350	Assessment of linkage disequilibrium patterns between structural variants and single nucleotide polymorphisms in three commercial chicken populations.. **2022**, 23, 193	1
349	Guidelines for bioinformatics of single-cell sequencing data analysis in Alzheimer's disease: review, recommendation, implementation and application.. **2022**, 17, 17	2
348	RF4Del: A Random Forest approach for accurate deletion detection.	
347	Fine-mapping and identification of candidate causal genes for tail length in the Merinolandschaf breed.	0
346	Evaluation of Genetic Structure of Pistachio Through Whole Genome Resequencing. 135-140	
345	A bulked segregant analysis tool for out-crossing species (BSATOS) and QTL-based genomics-assisted prediction of complex traits in apple. **2022**,	1
344	Comprehensive analysis of recessive carrier status using exome and genome sequencing data in 1543 Southern Chinese.. **2022**, 7, 23	1
343	Variation in spontaneous mutation rate and spectrum across the genome of Neurospora crassa.	0
342	Characterisation of the Function of a SINE-VNTR- Retrotransposon to Modulate Isoform Expression at the Locus.. **2022**, 15, 815695	0
341	Unravelling the tumour genome: the evolutionary and clinical impacts of structural variants in tumourigenesis.. **2022**,	0
340	Integrating whole genome sequencing, methylation, gene expression, topological associated domain information in regulatory mutation prediction: A study of follicular lymphoma.. **2022**, 20, 1726-1742	
339	Locus specific reduction of L1 expression in the cortices of individuals with amyotrophic lateral sclerosis.. **2022**, 15, 25	
338	svBreak: A New Approach for the Detection of Structural Variant Breakpoints Based on Convolutional Neural Network.. **2022**, 2022, 7196040	
337	Detecting structural variations with precise breakpoints using low-depth WGS data from a single oxford nanopore MinION flowcell.. **2022**, 12, 4519	1
336	Integrating whole-genome sequencing with multi-omic data reveals the impact of structural variants on gene regulation in the human brain.. **2022**,	1

335	Spatiotemporal variations in retrovirus-host interactions among Darwin finches.	
334	Elongin C (ELOC/TCEB1) associated von Hippel-Lindau disease.. **2022**,	0
333	A neurodegenerative disease landscape of rare mutations in Colombia due to founder effects.. **2022**, 14, 27	1
332	Accurate detection of structural variation is hard.	0
331	Single nucleotide polymorphisms (SNPs) and indels identified from whole-genome re-sequencing of four Chinese donkey breeds.. **2022**, 1-12	
330	Alterations in 3D chromatin organization contribute to tumorigenesis of -amplified glioblastoma.. **2022**, 20, 1967-1978	0
329	Genomic insights into present local adaptation and future climate change vulnerability of a keystone forest tree species in East Asian.	0
328	Novel structural variant genome detection in extended pedigrees through negative binomial optimization. **2021**,	
327	The SeqAn Library (Versions 1 and 2). **2022**, 7-32	
326	Full Issue PDF.. **2021**, 34, 1228-1333	
325	RB1 loss triggers dependence on ESRRG in retinoblastoma.	
324	Dosage sensitivity and exon shuffling shape the landscape of polymorphic duplicates in Drosophila and humans.. **2021**,	0
323	HolistIC: leveraging Hi-C and whole genome shotgun sequencing for double minute chromosome discovery. *Bioinformatics*, **2021**,	7.2
322	Whole genome sequencing reveals large deletions and other loss of function mutations in drug resistance genes. **2021**, 7,	0
321	CONGA: Copy number variation genotyping in ancient genomes and low-coverage sequencing data.	0
320	Hidden biases in germline structural variant detection.. **2021**, 22, 347	3
319	Computational analysis of cancer genome sequencing data. **2021**,	2
318	Diverse tumorigenic consequences of human papillomavirus integration in primary oropharyngeal cancers.. **2021**,	2

317	A Comparison of Tools for Copy-Number Variation Detection in Germline Whole Exome and Whole Genome Sequencing Data.. **2021**, 13,	1
316	GIP: an open-source computational pipeline for mapping genomic instability from protists to cancer cells.. **2021**,	0
315	ParaPindel: a scalable coordinated parallel detection framework for human genome-wide structural variation. **2021**,	
314	Whole-genome resequencing of worldwide wild and domestic sheep elucidates genetic diversity, introgression and agronomically important loci. **2021**,	4
313	Variant calling: considerations, practices, and developments. **2021**,	0
312	PocaCNV: A Tool to Detect Copy Number Variants from Population-Scale Genome Sequencing Data. **2021**,	
311	Integrated analysis of whole genome and transcriptome sequencing reveals a frameshift mutation associated with recessive embryonic lethality in Holstein cattle. **2021**,	0
310	Endogenous protein tagging in medaka using a simplified CRISPR/Cas9 knock-in approach. **2021**, 10,	4
309	An ensemble deep learning framework to refine large deletions in linked-reads. **2021**,	
308	Whole genome sequencing-based copy number variations reveal novel pathways and targets in Alzheimer's disease.. **2021**,	0
307	Sorghum Association Panel Whole-Genome Sequencing Establishes Pivotal Resource for Dissecting Genomic Diversity.	
306	[Application of high throughput sequencing in pediatric neurooncology].. **2022**, 84, 58-63	
305	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes.. **2022**,	1
304	Life-threatening viral disease in a novel form of autosomal recessive IFNAR2 deficiency in the Arctic.. **2022**, 219,	1
303	Genome assembly, structural variants, and genetic differentiation between Lake Whitefish young species pairs (Coregonus sp.) with long and short reads.. **2022**,	1
302	Microevolution of the and Reinforces the Bias of Quorum Sensing System in Laboratory Strains of PAO1.. **2022**, 13, 821895	1
301	Experimental evolution in TP53 deficient gastric organoids recapitulates tumorigenesis.	0
300	Genomic and transcriptomic analysis of a library of small cell lung cancer patient-derived xenografts.. **2022**, 13, 2144	0

299 Overview of structural variation calling: Simulation, identification, and visualization.. **2022**, 145, 105534 0

298 Data_Sheet_1.docx. **2020**,

297 Image_1.tif. **2019**,

296 Image_2.tif. **2019**,

295 Image_3.tif. **2019**,

294 Image_4.tif. **2019**,

293 Table_1.xlsx. **2019**,

292 Table_2.xlsx. **2019**,

291 Table_3.xlsx. **2019**,

290 Data_Sheet_1.docx. **2020**,

289 Data_Sheet_1.pdf. **2019**,

288 Image_1.TIF. **2019**,

287 Image_2.JPEG. **2019**,

286 Table_1.XLSX. **2019**,

285 Table_2.XLSX. **2019**,

284 Table_3.XLSX. **2019**,

283 Table_4.XLSX. **2019**,

282 Table_5.XLSX. **2019**,

281 Data_Sheet_1.pdf. **2018**,

280 Data_Sheet_2.ZIP. **2018**,

279 Data_Sheet_3.XLSX. **2018**,

278 Data_Sheet_4.XLSX. **2018**,

277 Data_Sheet_5.XLSX. **2018**,

276 Data_Sheet_6.XLSX. **2018**,

275 Data_Sheet_7.XLSX. **2018**,

274 Data_Sheet_8.XLSX. **2018**,

273 Data_Sheet_1.PDF. **2019**,

272 Data_Sheet_10.PDF. **2019**,

271 Data_Sheet_11.PDF. **2019**,

270 Data_Sheet_12.PDF. **2019**,

269 Data_Sheet_13.PDF. **2019**,

268 Data_Sheet_2.PDF. **2019**,

267 Data_Sheet_3.PDF. **2019**,

266 Data_Sheet_4.PDF. **2019**,

265 Data_Sheet_5.PDF. **2019**,

264 Data_Sheet_6.PDF. **2019**,

263 Data_Sheet_7.PDF. **2019**,

262 Data_Sheet_8.PDF. **2019**,

261 Data_Sheet_9.PDF. **2019**,

260 Table_1.DOCX. **2019**,

259 Table_2.DOCX. **2019**,

258 Table_3.xlsx. **2019**,

257 Table_4.xlsx. **2019**,

256 Data_Sheet_1.PDF. **2019**,

255 Table_1.DOCX. **2019**,

254 Table_2.DOCX. **2019**,

253 Image_1.png. **2020**,

252 Table_1.docx. **2020**,

251 Table_2.docx. **2020**,

250 Data_Sheet_1.XLSX. **2018**,

249 Presentation_1.PDF. **2018**,

248 Data_Sheet_1.docx. **2020**,

247 Data_Sheet_2.xls. **2020**,

246 Presentation_1.zip. **2019**,

245 Table_1.xlsx. **2019**,

244 Table_2.xlsx. **2019**,

243 Table_3.xlsx. **2019**,

242 Table_4.xlsx. **2019**,

241 Table_5.xlsx. **2019**,

240 DataSheet_1.docx. **2019**,

239 DataSheet1.ZIP. **2018**,

238 Image1.PDF. **2018**,

237 Image2.pdf. **2018**,

236 Image3.pdf. **2018**,

235 Table1.XLSX. **2018**,

234 Table2.XLSX. **2018**,

233 Table3.XLSX. **2018**,

232 Table4.XLSX. **2018**,

231 Structural variants in the barley gene pool: precision and sensitivity to detect them using short-read sequencing and their association with gene expression and phenotypic variation. 0

230 Somatic whole genome dynamics of precancer in Barrett's esophagus reveals features associated with disease progression.. **2022**, 13, 2300 0

229 A deep learning framework for structural variant discovery and genotyping.

228 A Simulator for Somatic Evolution Study Design. 1

227	Genome-wide analyses of introgression between two sympatric Asian oak species.. **2022**,	0
226	TT-Mars: structural variants assessment based on haplotype-resolved assemblies.. **2022**, 23, 110	1
225	Genomic patterns of malignant peripheral nerve sheath tumour (MPNST) evolution correlate with clinical outcome and are detectable in cell-free DNA.	
224	Capture Rate of V(D)J Sequencing for Minimal Residual Disease Detection in Multiple Myeloma.. **2022**, 28, 2160-2166	0
223	The third international hackathon for applying insights into large-scale genomic composition to use cases in a wide range of organisms. 11, 530	
222	Focal structural variants revealed by whole genome sequencing disrupt the histone demethylase in B cell lymphomas.. **2022**,	0
221	Molecular landscape and therapeutic alterations in Asian soft-tissue sarcoma patients.. **2022**,	
220	DNA sequence features underlying large-scale duplications and deletions in human.. **2022**,	
219	Genetic and immunohistochemical profiling of small cell and large cell neuroendocrine carcinomas of the breast.. **2022**,	0
218	Absence of increased genomic variants in the cyanobacterium Chroococcidiopsis exposed to Mars-like conditions outside the space station.. **2022**, 12, 8437	2
217	Whole-genome and transcriptome analysis of advanced adrenocortical cancer highlights multiple alterations affecting epigenome and DNA repair pathways.. **2022**, 8,	0
216	A two-step adaptive walk rewires nutrient transport in a challenging edaphic environment.. **2022**, 8, eabm93851	
215	Extensive mosaicism by somatic L1 retrotransposition in normal human cells.	
214	Population-scale genotyping of structural variation in the era of long-read sequencing. **2022**,	0
213	A comparative study of structural variant calling strategies using the Alzheimer Disease Sequencing Project whole genome family data.	
212	Whole genome sequencing identifies a missense polymorphism in PADI6 associated with testicular/ovotesticular XX disorder of sex development in dogs. **2022**, 114, 110389	0
211	Alterations in transcriptional networks in cancer: the role of noncoding somatic driver mutations. **2022**, 75, 101919	1
210	snakeSV: Flexible Framework for Large-Scale SV Discovery. **2022**, 1-14	

209	Transposable Element Structural Variants in Parkinson Disease: Focusing on Genotyping Alu Transposable Element Insertions with TypeTE. **2022**, 43-62	
208	Analysis of the Retrotransposon SINE-VNTR-Alu (SVA) Polymorphisms in the Genetics and Pathophysiology of Complex Diseases. **2022**, 63-77	
207	DNAscan2: a versatile, scalable, and user-friendly analysis pipeline for next-generation sequencing data.	
206	Genomic architecture of Autism Spectrum Disorder from comprehensive whole-genome sequence annotation.	0
205	Generation of hypoimmunogenic induced pluripotent stem cells by CRISPR-Cas9 system and detailed evaluation for clinical application.. **2022**,	0
204	Low spontaneous mutation rate in a complex multicellular eukaryote with a haploid-diploid life cycle.	0
203	Detection of Small CYP11B1 Deletions and One Founder Chimeric CYP11B2/CYP11B1 Gene in 11Hydroxylase Deficiency. **2022**, 13,	0
202	Sorghum Association Panel Whole-Genome Sequencing Establishes Cornerstone Resource for Dissecting Genomic Diversity.	1
201	Structural Variation in Cancer: Role, Prevalence, and Mechanisms. **2022**, 23,	0
200	Genetic and peripheral visual system changes underlie evolving butterfly mate preference.	
199	Expansion of a retrovirus lineage in the koala genome. **2022**, 119,	
198	Integrated genomic analyses of acral and mucosal melanomas nominate novel driver genes. **2022**, 14,	1
197	The Relative Power of Structural Genomic Variation versus SNPs in Explaining the Quantitative Trait Growth in the Marine Teleost Chrysophrys auratus. **2022**, 13, 1129	0
196	EagleC: A deep-learning framework for detecting a full range of structural variations from bulk and single-cell contact maps. **2022**, 8,	1
195	Transposable element-mediated rearrangements are prevalent in human genomes.	0
194	Dissection of a complex enhancer cluster at the Sox2 locus.	0
193	SARS-CoV-2 minor variant genomes at the start of the pandemic contained markers of VoCs.	
192	Mouse genomic rewriting and tailoring: synthetic Trp53 and humanized ACE2.	0

191	PhenGenVar: A User-Friendly Genetic Variant Detection and Visualization Tool for Precision Medicine. **2022**, 12, 959	1
190	Evolving copy number gains promote tumor expansion and bolster mutational diversification.	0
189	ClinCNV: multi-sample germline CNV detection in NGS data.	0
188	Meerkat: An Algorithm to Reliably Identify Structural Variations and Predict Their Forming Mechanisms. **2022**, 107-135	
187	High-quality genome assembly and pan-genome studies facilitate genetic discovery in mung bean and its improvement. **2022**, 100352	1
186	A comprehensive benchmarking of WGS-based deletion structural variant callers. **2022**, 23,	0
185	Consensus-Based Identification and Comparative Analysis of Structural Variants and Their Influence on 3D Genome Structure Using Long- and Short-Read Sequencing Technologies in Polish Families. **2023**, 41-49	
184	Maffucci syndrome complicated by giant chondrosarcoma in the left ankle with an IDH1 R132C mutation: a case report. **2022**, 20,	
183	Contrasting patterns of single nucleotide polymorphisms and structural variations across multiple invasions.	0
182	Pollen sequencing reveals barriers and aberrant patterns of recombination in interspecific tomato hybrids.	
181	3'RNA and whole-genome sequencing of archival uterine leiomyomas reveal a tumor subtype with chromosomal rearrangements affecting either HMGA2, HMGA1 , or PLAG1.	
180	No evidence of increased mutations in the germline of a group of British nuclear test veterans. **2022**, 12,	0
179	CanSeer: A Method for Development and Clinical Translation of Personalized Cancer Therapeutics.	
178	Experimental introgression in Drosophila: asymmetric postzygotic isolation associated with chromosomal inversions and an incompatibility locus on the X chromosome.	0
177	Genomic and epigenomic BRCA alterations predict adaptive resistance and response to platinum-based therapy in patients with triple-negative breast and ovarian carcinomas. **2022**, 14,	3
176	Patient-Specific Assays Based on Whole-Genome Sequencing Data to Measure Residual Disease in Children With Acute Lymphoblastic Leukemia: A Proof of Concept Study. 12,	
175	A molecular phenotypic map of Malignant Pleural Mesothelioma.	
174	Identification of a candidate sex determination gene in Culaea inconstans suggests convergent recruitment of an Amh duplicate in two lineages of stickleback.	2

173	Time Series Genomics of Pseudomonas aeruginosa Reveals the Emergence of a Hypermutator Phenotype and Within-Host Evolution in Clinical Inpatients.	
172	Heterozygotic Brca1 mutation initiates mouse genome instability at embryonic stage. **2022**, 11,	0
171	High frequency of genetic/epigenetic disorders in short stature children born with very low birth weight.	1
170	Portable sequencing of Mycobacterium tuberculosis for clinical and epidemiological applications.	0
169	Cloning and base editing of GFP transgenic rhesus monkey and off-target analysis. **2022**, 8,	0
168	Clinical impact of whole-genome sequencing in patients with early-onset dementia. jnnp-2021-328146	1
167	Centuries of genome instability and evolution in soft-shell clam transmissible cancer.	0
166	Assembly of a pangenome for global cattle reveals missing sequences and novel structural variations, providing new insights into their diversity and evolutionary history. **2022**, 32, 1585-1601	0
165	Recent, full-length gene retrocopies are common in canids. **2022**, 32, 1602-1611	1
164	Prediction of the 3D cancer genome from genomic rearrangements using InfoHiC.	
163	CRISPR-Cas9 editing induces Loss of Heterozygosity in the pathogenic yeast Candida parapsilosis.	
162	Molecular characterization reveals subclasses of 1q gain in intermediate-risk Wilms tumors.	
161	Deciphering the Evolutionary History of Complex Rearrangements in Head and Neck Cancer Patients Using Multi-Omic Approach.	
160	A composite strategy of genome-wide association study and copy number variation analysis for carcass traits in a Duroc pig population. **2022**, 23,	0
159	RB1 loss triggers dependence on ESRRG in retinoblastoma. **2022**, 8,	0
158	The importance of genome sequencing: unraveling SSBP1 variant missed by exome sequencing. 1-5	
157	A pan-Zea genome map for enhancing maize improvement. **2022**, 23,	1
156	Structural variants in the barley gene pool: precision and sensitivity to detect them using short-read sequencing and their association with gene expression and phenotypic variation.	0

155	Multi-omics analyses reveal MdMYB10 hypermethylation being responsible for a bud sport of apple fruit color.	2
154	PerSVade: personalized structural variant detection in any species of interest. **2022**, 23,	0
153	Detection of early seeding of Richter transformation in chronic lymphocytic leukemia. **2022**, 28, 1662-1671	1
152	Molecular Characterization of Adenocarcinomas Arising in the Urinary Bladder Following Augmentation Cystoplasty: A Multi-Institutional Study. **2022**,	0
151	Evolutionary genomics of structural variation in the tea plant, <i>Camellia sinensis</i>. **2022**, 1, 1-11	0
150	Simultaneous Mapping of Enhancers and Enhancer Rearrangements with Paired-End H3K27ac ChIP-seq. **2022**, 131-140	0
149	Tangent normalization for somatic copy-number inference in cancer genome analysis.	0
148	A genetic model for central chondrosarcoma evolution correlates with patient outcome. **2022**, 14,	1
147	A second unveiling: haplotig masking of the eastern oyster genome improves population-level inference.	0
146	MYRF: A New Regulator of Cardiac and Early Gonadal Development Insights from Single Cell RNA Sequencing Analysis. **2022**, 11, 4858	0
145	Systematic Analysis of Copy Number Variations in the Pathogenic Yeast Candida parapsilosis Identifies a Gene Amplification in RTA3 That is Associated with Drug Resistance.	1
144	Pangenomic analysis of Chinese gastric cancer. **2022**, 13,	0
143	Haplotype-resolved assemblies and variant benchmark of a Chinese Quartet.	0
142	A survey of current methods to detect and genotype inversions.	0
141	Fine-mapping and identification of candidate causal genes for tail length in the Merinolandschaf breed. **2022**, 5,	0
140	Structural variation and eQTL analysis in two experimental populations of chickens divergently selected for feather pecking behavior.	0
139	Genome-wide association study of traits in sacred lotus uncovers MITE-associated variants underlying stamen petaloid and petal number variations. 13,	1
138	Evolutionary Trajectories of Primary and Metastatic Pancreatic Neuroendocrine Tumors Based on Genomic Variations. **2022**, 13, 1588	0

137	Genomic evidence reveals intraspecific divergence of the hot-spring snake (Thermophis baileyi), an endangered reptile endemic to the Qinghai-Tibet plateau.	1
136	Structural variants and tandem repeats in the founder individuals of four F2 pig crosses and implications to F2 GWAS results. **2022**, 23,	0
135	Genome-wide detection of copy number variation in American mink using whole-genome sequencing. **2022**, 23,	0
134	Case Report: Consistent disease manifestations with a staggered time course in two identical twins affected by adenosine deaminase 2 deficiency. 13,	0
133	Improved pea reference genome and pan-genome highlight genomic features and evolutionary characteristics. **2022**, 54, 1553-1563	2
132	An oligogenic architecture underlying ecological and reproductive divergence in sympatric populations.	0
131	Nucleotide and structural polymorphisms of the eastern oyster genome paint a mosaic of divergence, selection, and human impacts.	0
130	The genomic landscape of pediatric acute lymphoblastic leukemia. **2022**, 54, 1376-1389	7
129	Disruption of the topologically associated domain at Xp21.2 is related to 46,XY gonadal dysgenesis. jmedgenet-2022-108	
128	Identification of region of difference and H37Rv-related deletion in Mycobacterium tuberculosis complex by structural variant detection and genome assembly. 13,	0
127	Whole Genome Sequencing Reveals the Structure of Environment-Associated Divergence in a Broadly Distributed Montane Bumble Bee, Bombus vancouverensis. **2022**, 6,	1
126	Quartet DNA reference materials and datasets for comprehensively evaluating germline variants calling performance.	0
125	Treatment-mediated selection of lethal prostate cancer clones defined by copy number architectures.	0
124	Next-generation phenotyping contributing to the identification of a 4.7 kb deletion in KANSL1 causing Koolen-de Vries syndrome.	0
123	CAFE (Cohort Allele Frequency Estimation) Pipeline: A workflow to generate a variant catalogue from Whole Genome Sequences.	0
122	Prevalence and mechanisms of somatic deletions in single human neurons during normal aging and in DNA repair disorders. **2022**, 13,	0
121	Reconstruct a eukaryotic chromosome arm by de novo design and synthesis.	0
120	Haplotype-resolved assemblies and variant benchmark of a Chinese Quartet.	0

119	Lynch Syndrome Identification in Saudi Cohort of Endometrial Cancer Patients Screened by Universal Approach. **2022**, 23, 12299	0
118	An Ultra-Rare Manifestation of an X-Linked Recessive Disorder: Duchenne Muscular Dystrophy in a Female Patient. **2022**, 23, 13076	0
117	Loss of altruism in the social amoebaDictyostelium discoideumis associated with the G protein-coupled receptorgrlG.	0
116	Approaches to long-read sequencing in a clinical setting to improve diagnostic rate. **2022**, 12,	1
115	The genomic basis of reproductive and migratory behaviour in a polymorphic salmonid.	0
114	Targeted profiling of human extrachromosomal DNA by CRISPR-CATCH.	2
113	Characterization of somatic structural variations in 528 Chinese individuals with Esophageal squamous cell carcinoma. **2022**, 13,	0
112	Leveraging patient derived models of FGFR2 fusion positive intrahepatic cholangiocarcinoma to identify synergistic therapies. **2022**, 6,	1
111	Spatiotemporal variations in retrovirus-host interactions among Darwin□ finches. **2022**, 13,	0
110	Subtype-specific 3D genome alteration in acute myeloid leukaemia.	1
109	Comprehensive Molecular Characterization of Gallbladder Carcinoma and Potential Targets for Intervention.	1
108	Characteristics and potential functional effects of long insertions in Asian butternuts. **2022**, 23,	0
107	Molecular Characterization Reveals Subclasses of 1q Gain in Intermediate Risk Wilms Tumors. **2022**, 14, 4872	0
106	How Genetics and Genomics Advances Are Rewriting Pediatric Cancer Research and Clinical Care. **2022**, 58, 1386	0
105	NGS-Based Molecular Karyotyping of Multiple Myeloma: Results from the GEM12 Clinical Trial. **2022**, 14, 5169	0
104	Integration of Hi-C with short and long-read genome sequencing reveals the structure of germline rearranged genomes. **2022**, 13,	0
103	Influences of rare copy-number variation on human complex traits. **2022**, 185, 4233-4248.e27	0
102	A□-□4.1□kb deletion in IRX1 gene upstream is completely associated with rumplessness in Piao chicken. **2022**, 114, 110515	0

101	Detailed analysis of mortality rates in the female progeny of 1,001 Holstein bulls allows the discovery of new dominant genetic defects. **2022**,	0
100	Severe inbreeding, increased mutation load and gene loss-of-function in the critically endangered Devils Hole pupfish. **2022**, 289,	2
99	Druggable Growth Dependencies and Tumor Evolution Analysis in Patient-Derived Organoids of Neuroendocrine Cancer.	0
98	Genomic insights into local adaptation and future climate-induced vulnerability of a keystone forest tree in East Asia. **2022**, 13,	1
97	Plasma Sequencing for Patients with GISTLimitations and Opportunities in an Academic Setting. **2022**, 14, 5496	0
96	Characterization of the genetic and regulatory networks associated with sugar and acid metabolism in apples via an integrated strategy. 13,	0
95	A rapid platform for 3D patient-derived cutaneous neurofibroma organoid establishment and screening.	0
94	Personalized genome assembly for accurate cancer somatic mutation discovery using tumor-normal paired reference samples. **2022**, 23,	0
93	Uterine Inflammatory Myofibroblastic Tumors. Publish Ahead of Print,	0
92	Landscape of potentially targetable receptor tyrosine kinase fusions in diverse cancers by DNA-based profiling. **2022**, 6,	0
91	Investigation of high gamma-glutamyltransferase syndrome in California Thoroughbred racehorses.	0
90	Whole-genome sequencing of chronic lymphocytic leukemia identifies subgroups with distinct biological and clinical features. **2022**, 54, 1675-1689	2
89	Genomic architecture of autism from comprehensive whole-genome sequence annotation. **2022**, 185, 4409-4427.e18	1
88	Transposable element-mediated rearrangements are prevalent in human genomes. **2022**, 13,	0
87	Ancestrally polymorphic chromosomal inversions as potential drivers of speciation.	0
86	Robust identification of extrachromosomal DNA and genetic variants using multiple genetic abnormality sequencing (MGA-Seq).	0
85	Structural variation and eQTL analysis in two experimental populations of chickens divergently selected for feather-pecking behavior.	0
84	TLsub: A transfer learning based enhancement to accurately detect mutations with wide-spectrum sub-clonal proportion. 13,	0

83	Return of non-ACMG recommended incidental genetic findings to pediatric patients: considerations and opportunities from experiences in genomic sequencing. **2022**, 14,	0
82	From Samples to Germline and Somatic Sequence Variation: A Focus on Next-Generation Sequencing in Melanoma Research. **2022**, 12, 1939	1
81	Very Low Rates of Spontaneous Gene Deletions and Gene Duplications in Dictyostelium discoideum.	0
80	Retrotransposon instability dominates the acquired mutation landscape of mouse induced pluripotent stem cells. **2022**, 13,	0
79	Experimental introgression in Drosophila : Asymmetric postzygotic isolation associated with chromosomal inversions and an incompatibility locus on the X chromosome.	0
78	Genomic heterogeneity as a barrier to precision oncology in urothelial cancer. **2022**, 41, 111859	0
77	Structural variant analysis of a cancer reference cell line sample using multiple sequencing technologies. **2022**, 23,	0
76	Novel correlative analysis identifies multiple genomic variations impacting ASD with macrocephaly.	0
75	Ultrafast prediction of somatic structural variations by filtering out reads matched to pan-genome k-mer sets.	0
74	Global dissection of the recombination landscape in soybean using a high-density 600K SoySNP array.	1
73	Optical Genome Mapping for detecting Homologous Recombination Deficiency (HRD) in human breast cancers.	0
72	Noninvasive prenatal screening for cystic fibrosis using circulating trophoblasts: Detection of the 50 most common disease-causing variants.	0
71	CRISPR-Cas9 Editing Induces Loss of Heterozygosity in the Pathogenic Yeast Candida parapsilosis. **2022**, 7,	0
70	CONGA: Copy number variation genotyping in ancient genomes and low-coverage sequencing data. **2022**, 18, e1010788	0
69	Systematic assessment of the contribution of structural variants to inherited retinal diseases.	1
68	Origin Matters: Using a Local Reference Genome Improves Measures in Population Genomics.	0
67	Case Report: Whole genome sequencing identifies CCDC88C as a novel JAK2 fusion partner in pediatric T-cell acute lymphoblastic leukemia. 10,	0
66	Genomic patterns of malignant peripheral nerve sheath tumor (MPNST) evolution correlate with clinical outcome and are detectable in cell-free DNA.	0

65	Genomic basis of Y-linked dwarfism in cichlids pursuing alternative reproductive tactics.	0
64	Integrated Genomic and Clinicopathologic Approach Distinguishes Pancreatic Grade 3 Neuroendocrine Tumor From Neuroendocrine Carcinoma and Identifies a Subset With Molecular Overlap. **2023**, 100065	0
63	Third-generation genome sequencing implicates medium-sized structural variants in chronic schizophrenia. 16,	0
62	Germline TP53 mutations undergo copy number gain years prior to tumor diagnosis. **2023**, 14,	0
61	Integrative genomic and transcriptomic analyses of a bud sport mutant □inzao Wuhe□with the phenotype of large berries in grapevines. 11, e14617	2
60	Profilin 1 deficiency drives mitotic defects and reduces genome stability. **2023**, 6,	0
59	Different structural variant prediction tools yield considerably different results in Caenorhabditis elegans. **2022**, 17, e0278424	0
58	561. Unravelling the structural variability in chicken genomes by long-read sequencing. **2022**,	0
57	Development and characterization of new patient-derived xenograft (PDX) models of osteosarcoma with distinct metastatic capacities.	0
56	Comprehensive analysis of mutational signatures reveals distinct patterns and molecular processes across 27 pediatric cancers.	0
55	Duck pan-genome reveals two transposon-derived structural variations caused bodyweight enlarging and white plumage phenotype formation during evolution.	0
54	An expectation□aximization algorithm for estimating proportions of deletions among bacterial populations with application to study antibiotic resistance gene transfer in Enterococcus faecalis.	0
53	Inversions maintain differences between migratory phenotypes of a songbird. **2023**, 14,	0
52	Heritable defects in telomere and mitotic function selectively predispose to sarcomas. **2023**, 379, 253-260	1
51	Detecting genomic deletions from high-throughput sequence data with unsupervised learning. **2022**, 23,	0
50	Structural variation across 138,134 samples in the TOPMed consortium.	0
49	Visualizing and exploring patterns of large mutational events with SigProfilerMatrixGenerator.	0
48	Rewiring of the 3D genome during acquisition of carboplatin resistance in a triple-negative breast cancer patient-derived xenograft. **2023**, 13,	0

47	Long-read sequencing of diagnosis and post-therapy medulloblastoma reveals complex rearrangement patterns and epigenetic signatures. **2023**, 3, 100281	0
46	SVsearcher: A more accurate structural variation detection method in long read data. **2023**, 158, 106843	0
45	Dynamic Genomes - Mechanisms and consequences of genomic diversity impacting plant-fungal interactions. **2023**, 125, 102006	0
44	A Clonal Evolution Simulator for Planning Somatic Evolution Studies. **2022**, 229-242	0
43	Next-Generation Sequencing (NGS) and Third-Generation Sequencing (TGS) for the Diagnosis of Thalassemia. **2023**, 13, 373	2
42	Benchmarking of structural variant detection in the tetraploid potato genome using linked-read sequencing. **2023**, 115, 110568	0
41	Ultra-Low Level Somatic Mutations and Structural Variations in Focal Cortical Dysplasia Type II.	0
40	A molecular phenotypic map of malignant pleural mesothelioma. **2022**, 12,	0
39	Clonal hematopoiesis driven by chromosome 1q/MDM4 trisomy defines a canonical route toward leukemia in Fanconi anemia. **2023**, 30, 153-170.e9	2
38	Somatic nuclear mitochondrial DNA insertions are prevalent in the human brain and accumulate in aging fibroblasts.	0
37	BARD1germline variants induce haploinsufficiency and DNA repair defects in neuroblastoma.	0
36	Genomic architecture underlying morphological and physiological adaptation to high elevation in a songbird.	0
35	Development of homozygous tetraploid potato and whole genome doubling-induced the enrichment of H3K27ac and potentially enhanced resistance to cold-induced sweetening in tubers. **2023**, 10,	0
34	Contributions of rare and common variation to early-onset and atypical dementia risk.	0
33	Insights into the convergent evolution of fructan biosynthesis in angiosperms from the highly characteristic chicory genome. **2023**, 238, 1245-1262	0
32	390. Integrating imputed structural variants and tandem repeats in a GWAS for growth and carcass traits in F2 pig crosses. **2022**,	0
31	265. Unravelling the effect of structural variants from whole-genome sequence for depigmentation in Nellore cattle. **2022**,	0
30	An allele-resolved nanopore-guided tour of the human placental methylome.	0

29	Distinct genomic contexts predict gene presence-absence variation in different pathotypes of a fungal plant pathogen.	0
28	Whole-Genome Comparison Reveals Structural Variations behind Heading Leaf Trait in Brassica oleracea. **2023**, 24, 4063	0
27	Systematic assessment of the contribution of structural variants to inherited retinal diseases.	0
26	Contrasting Patterns of Single Nucleotide Polymorphisms and Structural Variation Across Multiple Invasions. **2023**, 40,	0
25	Enhancer co-amplification and hijacking promote oncogene expression in liposarcoma.	0
24	Novel Exon 7 Deletions in TSPAN12 in a Three-Generation FEVR Family: A Case Report and Literature Review. **2023**, 14, 587	0
23	An oligogenic architecture underlying ecological and reproductive divergence in sympatric populations. 12,	0
22	Whole-genome sequencing reveals a complex African population demographic history and signatures of local adaptation. **2023**, 186, 923-939.e14	1
21	Whole-genome sequencing reveals an association between small genomic deletions and an increased risk of developing Parkinson□ disease. **2023**, 55, 555-564	0
20	INSnet: a method for detecting insertions based on deep learning network. **2023**, 24,	0
19	Genomic analysis of Leishmania turanica strains from different regions of Central Asia. **2023**, 17, e0011145	0
18	Impact and characterization of serial structural variations across humans and great apes.	0
17	The promise and challenges of characterizing genome-wide structural variants: A case study in a critically endangered parrot.	0
16	Synthetic regulatory genomics uncovers enhancer context dependence at the Sox2 locus. **2023**, 83, 1140-1152.e7	0
15	Multiomic analysis of malignant pleural mesothelioma identifies molecular axes and specialized tumor profiles driving intertumor heterogeneity. **2023**, 55, 607-618	0
14	The genomic characteristics affect phenotypic diversity from the perspective of genetic improvement of economic traits. **2023**, 26, 106426	0
13	Evidence for a single, ancient origin of a genus-wide alternative life history strategy. **2023**, 9,	0
12	Cue: a deep-learning framework for structural variant discovery and genotyping. **2023**, 20, 559-568	0

11	Homologous recombination deficiency signatures in gastrointestinal and thoracic cancers correlate with platinum therapy duration. **2023**, 7,	0
10	Integrative multi-omic sequencing reveals the MMTV-Myc mouse model mimics human breast cancer heterogeneity.	0
9	Cytosine base editors induce off-target mutations and adverse phenotypic effects in transgenic mice. **2023**, 14,	0
8	DNAscan2: a versatile, scalable, and user-friendly analysis pipeline for human next-generation sequencing data. **2023**, 39,	0
7	Test development, optimization and validation of a WGS pipeline for genetic disorders. **2023**, 16,	0
6	Resolution of structural variation in diverse mouse genomes reveals chromatin remodeling due to transposable elements. **2023**, 100291	0
5	Identification of copy number variations in the genome of Dairy Gir cattle. **2023**, 18, e0284085	0
4	Smart breeding driven by advances in sequencing technology. **2023**, 1, 43-56	1
3	3D genome mapping identifies subgroup-specific chromosome conformations and tumor-dependency genes in ependymoma. **2023**, 14,	0
2	Analysis of genome instability and implications for the consequent phenotype in Plasmodium falciparum containing mutated MSH2-1 (P513T). **2023**, 9,	0
1	A second unveiling: Haplotig masking of the eastern oyster genome improves population-level inference.	0