Bruno Dallapiccola

List of Publications by Year in Descending Order

560 papers	29,539 citations	78 h-index	152 g-index
572 ext. papers	33,250 ext. citations	5.9 avg, IF	6.22 L-index

#	Paper	IF	Citations
560	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. *Genes*, **2022**, 13, 889	4.2	0
559	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2021**, 108, 2112-2129	11	2
558	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
557	Cross-correlation of virome-bacteriome-host-metabolome to study respiratory health. *Trends in Microbiology*, **2021**,	12.4	4
556	Copy number variation analysis implicates novel pathways in patients with oculo-auriculo-vertebral-spectrum and congenital heart defects. *Clinical Genetics*, **2021**, 100, 268-279	4	2
555	Functional Genomics for Undiagnosed Patients: The Impact of Small GTPases Signaling Dysregulation at Pan-Embryo Developmental Scale. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 642235	5.7	0
554	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. *American Journal of Human Genetics*, **2021**, 108, 115-133	11	8
553	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
552	Clinical Application of Easychip 8x15K Platform in 4106 Pregnancies Without Ultrasound Anomalies. *Reproductive Sciences*, **2021**, 28, 1142-1149	3	0
551	Relationship between glucose homeostasis and obesity in early life-A study of Italian children and adolescents. *Human Molecular Genetics*, **2021**,	5.6	3
550	Homozygous and Gene Variants in a Boy with Growth Hormone Deficiency and Early Onset Osteoporosis. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
549	Fused Omics Data Models Reveal Gut Microbiome Signatures Specific of Inactive Stage of Juvenile Idiopathic Arthritis in Pediatric Patients. *Microorganisms*, **2020**, 8,	4.9	1
548	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. *Birth Defects Research*, **2020**, 112, 725-731	2.9	6
547	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. *BMC Pediatrics*, **2020**, 20, 120	2.6	3
546	The splice c.1815G>A variant in KIAA0586 results in a phenotype bridging short-rib-polydactyly and oral-facial-digital syndrome: A case report and literature review. *Medicine (United States)*, **2020**, 99, e19169	1.8	3
545	KBG syndrome: Common and uncommon clinical features based on 31 new patients. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1073-1083	2.5	10
544	Deficiency of MFSD7c results in microcephaly-associated vasculopathy in Fowler syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4081-4093	15.9	4

543	Skeletal abnormalities are common features in Aym□Gripp syndrome. *Clinical Genetics*, **2020**, 97, 362-369	4	4
542	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
541	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. *American Journal of Human Genetics*, **2020**, 106, 26-40	11	24
540	Delayed appearance of 3-methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. *American Journal of Medical Genetics, Part A*, **2020**, 182, 64-70	2.5	2
539	PPP1R21-related syndromic intellectual disability: Report of an adult patient and review. *American Journal of Medical Genetics, Part A*, **2020**, 182, 3014-3022	2.5	2
538	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
537	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *American Journal of Human Genetics*, **2019**, 105, 493-508	11	30
536	Expanding the clinical spectrum associated with PACS2 mutations. *Clinical Genetics*, **2019**, 95, 525-531	4	10
535	Familial aggregation of "apple peel" intestinal atresia and cardiac left-sided obstructive lesions: A possible causal relationship with NOTCH1 gene mutations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1570-1574	2.5	2
534	A familial chromosomal complex rearrangement confirms RUNX1T1 as a causative gene for intellectual disability and suggests that 1p22.1p21.3 duplication is likely benign. *Molecular Cytogenetics*, **2019**, 12, 26	2	
533	A heterozygous, intragenic deletion of CNOT2 recapitulates the phenotype of 12q15 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1615-1621	2.5	5
532	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. *New Genetics and Society*, **2019**, 38, 249-263	1.9	2
531	Association of Bright Liver With the PNPLA3 I148M Gene Variant in 1-Year-Old Toddlers. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2163-2170	5.6	4
530	Parent-of-Origin Effects in 15q11.2 BP1-BP2 Microdeletion (Burnside-Butler) Syndrome. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	14
529	Duplications of GPC3 and GPC4 genes in symptomatic female carriers of Simpson-Golabi-Behmel syndrome type 1. *European Journal of Medical Genetics*, **2019**, 62, 243-247	2.6	9
528	SOS1 mutations in Noonan syndrome: Cardiomyopathies and not only congenital heart defects! Report of six patients including two novel variants and literature review. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2083-2090	2.5	5
527	Potential of multiomics technology in precision medicine. *Current Opinion in Gastroenterology*, **2019** , 35, 491-498	3	13
526	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. *European Journal of Medical Genetics*, **2019**, 62, 103534	2.6	6

525	Microbiome Analytics of the Gut Microbiota in Patients With Juvenile Idiopathic Arthritis: A Longitudinal Observational Cohort Study. *Arthritis and Rheumatology*, **2019**, 71, 1000-1010	9.5	30
524	Confirmation of BRD4 haploinsufficiency role in Cornelia de Lange-like phenotype and delineation of a 19p13.12p13.11 gene contiguous syndrome. *Annals of Human Genetics*, **2019**, 83, 100-109	2.2	7
523	First case of nonalcoholic steatohepatitis in a child with del(1p36) and dup (Xp22): review of the literature. *Clinical Dysmorphology*, **2018**, 27, 42-45	0.9	2
522	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the diseaseN mechanism. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 446-451	2.3	7
521	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 397-405	3.5	10
520	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
519	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *Data in Brief*, **2018**, 16, 649-654	1.2	5
518	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. *Clinical Genetics*, **2018**, 93, 401-407	4	16
517	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 103, 305-316	11	21
516	Long-term survival and phenotypic spectrum in heterotaxy syndrome: A 25-year follow-up experience. *International Journal of Cardiology*, **2018**, 268, 100-105	3.2	17
515	Gut Microbiota Profiling and Gut-Brain Crosstalk in Children Affected by Pediatric Acute-Onset Neuropsychiatric Syndrome and Pediatric Autoimmune Neuropsychiatric Disorders Associated With Streptococcal Infections. *Frontiers in Microbiology*, **2018**, 9, 675	5.7	51
514	Gut Microbiota Markers in Obese Adolescent and Adult Patients: Age-Dependent Differential Patterns. *Frontiers in Microbiology*, **2018**, 9, 1210	5.7	78
513	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. *Human Mutation*, **2018**, 39, 1428-1441	4.7	10
512	Gut microbiota signatures in cystic fibrosis: Loss of host CFTR function drives the microbiota enterophenotype. *PLoS ONE*, **2018**, 13, e0208171	3.7	47
511	An additional patient with a homozygous mutation in DCPS contributes to the delination of Al-Raqad syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2781-2786	2.5	7
510	Small 4p16.3 deletions: Three additional patients and review of the literature. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2501-2508	2.5	7
509	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
508	First Report of Low-Rate Mosaicism for 20q11.21q12 Deletion and Delineation of the Associated Disorder. *Cytogenetic and Genome Research*, **2018**,	1.9	1

507	Gut microbiota profiling of pediatric nonalcoholic fatty liver disease and obese patients unveiled by an integrated meta-omics-based approach. *Hepatology*, **2017**, 65, 451-464	11.2	354
506	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. *Human Mutation*, **2017**, 38, 451-459	4.7	32
505	Reassessment of the 12q15 deletion syndrome critical region. *European Journal of Medical Genetics*, **2017**, 60, 220-223	2.6	7
504	Unclassifiable pattern of hypopigmentation in a patient with mosaic partial 12p tetrasomy without Pallister-Killian syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1943-1946	2.5	3
503	Expanding the phenotypic spectrum of truncating POGZ mutations: Association with CNS malformations, skeletal abnormalities, and distinctive facial dysmorphism. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1965-1969	2.5	17
502	Percentiles of serum uric acid and cardiometabolic abnormalities in obese Italian children and adolescents. *Italian Journal of Pediatrics*, **2017**, 43, 3	3.2	13
501	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
500	Interstitial 10q21.1q23.31 Duplication due to Meiotic Recombination of a Paternal Balanced Complex Rearrangement: Cytogenetic and Molecular Characterization. *Cytogenetic and Genome Research*, **2017**, 151, 179-185	1.9	1
499	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
498	Clinical and Neurobehavioral Features of Three Novel Kabuki Syndrome Patients with Mosaic KMT2D Mutations and a Review of Literature. *International Journal of Molecular Sciences*, **2017**, 19,	6.3	10
497	Congenital heart defects in molecularly proven Kabuki syndrome patients. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2912-2922	2.5	37
496	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *International Journal of Cardiology*, **2017**, 245, 92-98	3.2	48
495	-related intellectual disability syndrome: a recognisable entity. *Journal of Medical Genetics*, **2017**, 54, 613-623	5.8	28
494	"Omic" investigations of protozoa and worms for a deeper understanding of the human gut "parasitome". *PLoS Neglected Tropical Diseases*, **2017**, 11, e0005916	4.8	22
493	Reference ranges of HOMA-IR in normal-weight and obese young Caucasians. *Acta Diabetologica*, **2016**, 53, 251-60	3.9	98
492	Gut Microbiota Dysbiosis as Risk and Premorbid Factors of IBD and IBS Along the Childhood-Adulthood Transition. *Inflammatory Bowel Diseases*, **2016**, 22, 487-504	4.5	69
491	Survey of medical genetic services in Italy: year 2011. *BMC Health Services Research*, **2016**, 16, 96	2.9	2
490	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31

489	POPDC1(S201F) causes muscular dystrophy and arrhythmia by affecting protein trafficking. *Journal of Clinical Investigation*, **2016**, 126, 239-53	15.9	55
488	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. *Genetics and Molecular Biology*, **2016**, 39, 349-57	2	3
487	Monitoring Perinatal Gut Microbiota in Mouse Models by Mass Spectrometry Approaches: Parental Genetic Background and Breastfeeding Effects. *Frontiers in Microbiology*, **2016**, 7, 1523	5.7	10
486	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Velo-Cardio-Facial) syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 661-4	2.5	3
485	Foodomics as part of the host-microbiota-exposome interplay. *Journal of Proteomics*, **2016**, 147, 3-20	3.9	37
484	Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 962-973	11	55
483	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
482	Transmembrane 6 superfamily member 2 gene variant disentangles nonalcoholic steatohepatitis from cardiovascular disease. *Hepatology*, **2015**, 61, 506-14	11.2	311
481	BRF1 mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. *Genome Research*, **2015**, 25, 155-66	9.7	68
480	A de novo proximal 3q29 chromosome microduplication in a patient with oculo auriculo vertebral spectrum. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 797-801	2.5	18
479	Left ventricular non compaction with aortic valve anomalies: A recurrent feature of 22q11.2 distal deletion syndrome. *European Journal of Medical Genetics*, **2015**, 58, 406-8	2.6	1
478	AuthorsN response to J Thomas: "If not parthenogenesis why not in vivo embryogenesisNwith Mary as a birth mother". *Journal of Maternal-Fetal and Neonatal Medicine*, **2015**, 28, 1917	2	1
477	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. *American Journal of Human Genetics*, **2015**, 97, 99-110	11	48
476	Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the recruitment of the EvC complex and SMO into the cilium. *Human Molecular Genetics*, **2015**, 24, 4126-37	5.6	37
475	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 96, 753-64	11	54
474	Molecular Diversity and Associated Phenotypic Spectrum of Germline CBL Mutations. *Human Mutation*, **2015**, 36, 787-96	4.7	22
473	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
472	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. *European Journal of Human Genetics*, **2015**, 23, 1068-71	5.3	89

471	Connexin 26 variant carriers have a better gastrointestinal health: is this the heterozygote advantage?. *European Journal of Human Genetics*, **2015**, 23, 563-564	5.3	7
470	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. *Archives of Disease in Childhood*, **2015**, 100, 158-64	2.2	57
469	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
468	CHARGE syndrome due to deletion of region upstream of CHD7 gene START codon. *BMC Medical Genetics*, **2015**, 16, 78	2.1	5
467	Dysmorphologic assessment in 115 Mayer-Rokitansky-K□ter-Hauser patients. *Clinical Dysmorphology*, **2015**, 24, 95-101	0.9	6
466	Cardiovascular malformations in Adams-Oliver syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1175-7	2.5	10
465	Understanding probioticsN role in allergic children: the clue of gut microbiota profiling. *Current Opinion in Allergy and Clinical Immunology*, **2015**, 15, 495-503	3.3	14
464	Behavioral phenotype in Costello syndrome with atypical mutation: a case report. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 66-71	3.5	5
463	Phylogenetic and Metabolic Tracking of Gut Microbiota during Perinatal Development. *PLoS ONE*, **2015**, 10, e0137347	3.7	63
462	The Challenge of Prenatal Diagnostic Work-Up of Maternally Inherited X-Linked Opitz G/BBB: Case Report and Literature Review. *Case Reports in Obstetrics and Gynecology*, **2015**, 2015, 830108	0.8	2
461	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. *Circulation: Cardiovascular Genetics*, **2015**, 8, 572-581		65
460	Can modern biology interpret the mystery of the birth of Christ?. *Journal of Maternal-Fetal and Neonatal Medicine*, **2015**, 28, 240-4	2	7
459	Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K+ channel encoded by KCNJ6. *American Journal of Human Genetics*, **2015**, 96, 295-300	11	72
458	Hypoplastic left heart syndrome and 21q22.3 deletion. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 579-86	2.5	7
457	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. *BMC Medical Genetics*, **2014**, 15, 14	2.1	48
456	Identification of TBX5 mutations in a series of 94 patients with Tetralogy of Fallot. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3100-7	2.5	37
455	The human gut microbiota: a dynamic interplay with the host from birth to senescence settled during childhood. *Pediatric Research*, **2014**, 76, 2-10	3.2	144
454	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. *Human Genetics*, **2014**, 133, 1161-7	6.3	95

453	3p25.3 microdeletion of GABA transporters SLC6A1 and SLC6A11 results in intellectual disability, epilepsy and stereotypic behavior. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3061-8	2.5	21
452	A 4-polymorphism risk score predicts steatohepatitis in children with nonalcoholic fatty liver disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 632-6	2.8	58
451	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. *Human Molecular Genetics*, **2014**, 23, 4315-27	5.6	95
450	Meta-omic platforms to assist in the understanding of NAFLD gut microbiota alterations: tools and applications. *International Journal of Molecular Sciences*, **2014**, 15, 684-711	6.3	21
449	Nectin-4 mutations causing ectodermal dysplasia with syndactyly perturb the rac1 pathway and the kinetics of adherens junction formation. *Journal of Investigative Dermatology*, **2014**, 134, 2146-2153	4.3	24
448	Hypopigmented skin patches in 17q21.31 microdeletion syndrome: expanding the spectrum of cutaneous findings. *Clinical Dysmorphology*, **2014**, 23, 32-34	0.9	3
447	Mediterranean diet and health: food effects on gut microbiota and disease control. *International Journal of Molecular Sciences*, **2014**, 15, 11678-99	6.3	107
446	Genetics of Ebstein Anomaly **2014**, 25-30		1
445	The internet user profile of Italian families of patients with rare diseases: a web survey. *Orphanet Journal of Rare Diseases*, **2013**, 8, 76	4.2	39
444	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. *Orphanet Journal of Rare Diseases*, **2013**, 8, 75	4.2	15
443	Joubert syndrome and related disorders. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 113, 1879-88	3	60
442	The policy of public health genomics in Italy. *Health Policy*, **2013**, 110, 214-9	3.2	21
441	JAG1 mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3133-6	2.5	8
440	Exome sequencing in a family with intellectual disability, early onset spasticity, and cerebellar atrophy detects a novel mutation in EXOSC3. *Neurogenetics*, **2013**, 14, 247-50	3	31
439	SERCA1 protein expression in muscle of patients with Brody disease and Brody syndrome and in cultured human muscle fibers. *Molecular Genetics and Metabolism*, **2013**, 110, 162-9	3.7	20
438	De novo mutations of the gene encoding the histone acetyltransferase KAT6B in two patients with Say-Barber/Biesecker/Young-Simpson syndrome. *American Journal of Medical Genetics, Part A*, **2013** , 161A, 884-8	2.5	29
437	Congenital heart defects in recurrent reciprocal 1q21.1 deletion and duplication syndromes: rare association with pulmonary valve stenosis. *European Journal of Medical Genetics*, **2013**, 56, 144-9	2.6	27
436	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dyostosis. *European Journal of Medical Genetics*, **2013**, 56, 80-7	2.6	48

435	Atrioventricular canal defect in patients with RASopathies. *European Journal of Human Genetics*, **2013**, 21, 200-4	5.3	19
434	Clinical utility gene card for: Joubert syndrome--update 2013. *European Journal of Human Genetics*, **2013**, 21,	5.3	16
433	Association of DiGeorge anomaly and caudal dysplasia sequence in a neonate born to a diabetic mother. *Cardiology in the Young*, **2013**, 23, 14-7	1	5
432	Ablepharon macrostomia syndrome: A distinct genetic entity clinically related to the group of FRAS-FREM complex disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3012-7	2.5	6
431	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. *European Journal of Human Genetics*, **2013**, 21, 69-75	5.3	27
430	Overt cleft palate phenotype and TBX1 genotype correlations in velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2781-7	2.5	19
429	Early-life gut microbiota under physiological and pathological conditions: the central role of combined meta-omics-based approaches. *Journal of Proteomics*, **2012**, 75, 4580-7	3.9	48
428	Genetics of pediatric obesity. *Pediatrics*, **2012**, 130, 123-33	7.4	58
427	Deficiency for the ubiquitin ligase UBE3B in a blepharophimosis-ptosis-intellectual-disability syndrome. *American Journal of Human Genetics*, **2012**, 91, 998-1010	11	62
426	Kinematic and diffusion tensor imaging definition of familial Marcus Gunn jaw-winking synkinesis. *PLoS ONE*, **2012**, 7, e51749	3.7	12
425	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. *Human Mutation*, **2012**, 33, 1175-81	4.7	63
424	Nablus mask-like facial syndrome: deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2091-9	2.5	8
423	Atrioventricular canal defect as a sign of laterality defect in Ellis-van Creveld and polydactyly syndromes with ciliary and Hedgehog signaling dysfunction. *Pediatric Cardiology*, **2012**, 33, 874-5	2.1	10
422	Deletion of KDM6A, a histone demethylase interacting with MLL2, in three patients with Kabuki syndrome. *American Journal of Human Genetics*, **2012**, 90, 119-24	11	258
421	A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies Myhre syndrome. *American Journal of Human Genetics*, **2012**, 90, 161-9	11	64
420	Clinical utility gene card for: Mayer-Rokitansky-K□ter-Hauser syndrome. *European Journal of Human Genetics*, **2012**, 20,	5.3	17
419	RDDR: a dysmorphology diagnostic network for newborns in central Italy. *Journal of Maternal-Fetal and Neonatal Medicine*, **2012**, 25 Suppl 4, 121-3	2	1
418	Search of somatic GATA4 and NKX2.5 gene mutations in sporadic septal heart defects. *European Journal of Medical Genetics*, **2011**, 54, 306-9	2.6	31

417	Application of MLPA assay to characterize unsolved Eglobin gene rearrangements. *Blood Cells, Molecules, and Diseases*, **2011**, 46, 139-44	2.1	37
416	Atrioventricular canal defect and associated genetic disorders: new insights into polydactyly syndromes. *Neurology International*, **2011**, 1, 7	0	6
415	Low-rate repetitive nerve stimulation protocol in an Italian cohort of patients affected by recessive myotonia congenita. *Journal of Clinical Neurophysiology*, **2011**, 28, 39-44	2.2	17
414	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. *Nature Genetics*, **2011**, 43, 197-203	36.3	190
413	Copy-number variations involving the IHH locus are associated with syndactyly and craniosynostosis. *American Journal of Human Genetics*, **2011**, 88, 70-5	11	76
412	Mutations in ANKRD11 cause KBG syndrome, characterized by intellectual disability, skeletal malformations, and macrodontia. *American Journal of Human Genetics*, **2011**, 89, 289-94	11	161
411	Infantile cortical hyperostosis and COL1A1 mutation in four generations. *European Journal of Pediatrics*, **2011**, 170, 1385-90	4.1	11
410	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. *Neurogenetics*, **2011**, 12, 233-40	3	6
409	FGF17, a gene involved in cerebellar development, is downregulated in a patient with Dandy-Walker malformation carrying a de novo 8p deletion. *Neurogenetics*, **2011**, 12, 241-5	3	17
408	Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients. *Orphanet Journal of Rare Diseases*, **2011**, 6, 38	4.2	68
407	The difficult nosology of blepharophimosis-mental retardation syndromes: report on two siblings. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 459-65	2.5	12
406	Genetic dosage compensation in a family with velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 548-54	2.5	8
405	Deletion 2p15-16.1 syndrome: case report and review. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2473-8	2.5	23
404	The Pitt-Hopkins syndrome: report of 16 new patients and clinical diagnostic criteria. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1536-45	2.5	46
403	Ebstein anomaly: Genetic heterogeneity and association with microdeletions 1p36 and 8p23.1. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2196-202	2.5	28
402	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. *Human Mutation*, **2011**, 32, 760-72	4.7	82
401	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *Human Mutation*, **2011**, 32, 1278-89	4.7	48
400	Screening for 22q11.2 microdeletion in adults with tetralogy of Fallot. *Heart*, **2011**, 97, 860; author reply 860	5.1	

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