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The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies

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#	Paper IF	Citations
320	A new mouse model for the trisomy of the Abcg1-U2af1 region reveals the complexity of the combinatorial genetic code of down syndrome. 2009 , 18, 4756-69	86
319	A meta-commentary on the proposal for a meta-structure for DSM-V and ICD-11. 2009 , 39, 2099-103	13
318	Down syndrome: the crucible for treating genomic imbalance. 2009 , 11, 617-9	3
317	Down syndrome: comments and reflections on the 50th anniversary of Lejeune's discovery. 2009 , 149A, 2647-54	20
316	Trisomy of Erg is required for myeloproliferation in a mouse model of Down syndrome. 2010 , 115, 3966-9	53
315	The Erg-onomics of myeloproliferation in Down syndrome. 2010 , 115, 3859-60	
314	Achalasia: will genetic studies provide insights?. 2010 , 128, 353-64	70
313	Implications of copy number variation in people with chromosomal abnormalities: potential for greater variation in copy number state may contribute to variability of phenotype. 2010 , 4, 1-9	7
312	Implications of copy number variation in people with chromosomal abnormalities: potential for greater variation in copy number state may contribute to variability of phenotype. 2010 , 4, 1-9	9
311	The role of chromosome 21 in hematology and oncology. 2010 , 49, 497-508	23
310	Identification of a 21q22 duplication in a Silver-Russell syndrome patient further narrows down the Down syndrome critical region. 2010 , 152A, 356-9	12
309	Molecular diagnosis of Down syndrome using quantitative APEX-2 microarrays. 2010 , 30, 1170-7	1
308	Trisomic dose of several chromosome 21 genes perturbs haematopoietic stem and progenitor cell differentiation in Down's syndrome. 2010 , 29, 6102-14	38
307	Nucleotide-resolution analysis of structural variants using BreakSeq and a breakpoint library. 2010 , 28, 47-55	136
306	[Trisomy 21: fifty years between medicine and science]. 2010 , 26, 267-72	4
305	The clinical context of copy number variation in the human genome. 2010 , 12, e8	134
304	Classification of genome-wide copy number variations and their associated SNP and gene networks analysis. 2010 ,	

(2011-2010)

303	Down syndrome: from understanding the neurobiology to therapy. 2010 , 30, 14943-5		89
302	Cardiovascular Disorders among Persons with Down Syndrome. 2010 , 39, 165-194		1
301	Down's syndrome-like cardiac developmental defects in embryos of the transchromosomic Tc1 mouse. 2010 , 88, 287-95		47
300	Adults with genetic syndromes. 2010 , 9,		
299	Development of a novel selective inhibitor of the Down syndrome-related kinase Dyrk1A. 2010 , 1, 86		180
298	GABAB-GIRK2-mediated signaling in Down syndrome. 2010 , 58, 397-426		39
297	Molecular basis of pharmacotherapies for cognition in Down syndrome. 2010 , 31, 66-73		57
296	Down syndrome and the molecular pathogenesis resulting from trisomy of human chromosome 21. 2010 , 24, 87-99		8
295	[Progress in pediatric cardiology, congenital heart disease in adults, and heart surgery for congenital heart disease]. 2010 , 63 Suppl 1, 29-39		12
294	A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. 2010 , 11, R64		12
293	Phenotypic consequences of aneuploidy in Arabidopsis thaliana. 2010 , 186, 1231-45		75
292	What can we learn from study of Alzheimer's disease in patients with Down syndrome for early-onset Alzheimer's disease in the general population?. 2011 , 3, 13		11
291	MicroRNA-15a and -16-1 act via MYB to elevate fetal hemoglobin expression in human trisomy 13. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011 , 108, 1519-24	11.5	165
290	Down Syndrome. 2011 ,		3
289	The telomeric part of the human chromosome 21 from Cstb to Prmt2 is not necessary for the locomotor and short-term memory deficits observed in the Tc1 mouse model of Down syndrome. 2011 , 217, 271-81		27
288	Form of dual-specificity tyrosine-(Y)-phosphorylation-regulated kinase 1A nonphosphorylated at tyrosine 145 and 147 is enriched in the nuclei of astroglial cells, adult hippocampal progenitors, and some cholinergic axon terminals. 2011 , 195, 112-27		14
287	Brain plasticity and environmental enrichment in Ts65Dn mice, an animal model for Down syndrome. 71-84		3
286	Genetics of Down Syndrome. 2011 ,		3

285 Down Syndrome: A Complex and Interactive Genetic Disorder. 2011,

284	Myeloid leukemia in Down syndrome. 2011 , 16, 25-36	68
283	Exploratory Investigation on Functional Significance of ETS2 and SIM2 Genes in Down Syndrome. 2011 , 31, 247-257	5
282	Massive-scale RNA-Seq analysis of non ribosomal transcriptome in human trisomy 21. 2011 , 6, e18493	53
281	MNB/DYRK1A as a multiple regulator of neuronal development. 2011 , 278, 223-35	141
280	Decreased AIRE expression and global thymic hypofunction in Down syndrome. 2011 , 187, 3422-30	50
279	Educational paper: syndromic forms of primary immunodeficiency. 2011 , 170, 295-308	9
278	Genetic analysis of Down syndrome-associated heart defects in mice. 2011 , 130, 623-32	40
277	SNP and gene networks construction and analysis from classification of copy number variations data. 2011 , 12 Suppl 5, S4	10
276	Assembly of non-unique insertion content using next-generation sequencing. 2011 , 12 Suppl 6, S3	9
275	Meta-analysis of heterogeneous Down Syndrome data reveals consistent genome-wide dosage effects related to neurological processes. 2011 , 12, 229	78
274	Clinical manifestations of the deletion of Down syndrome critical region including DYRK1A and KCNJ6. 2011 , 155A, 113-9	43
273	Linking chromosome abnormality and copy number variation. 2011 , 155A, 469-75	7
272	Mental retardation, congenital heart malformation, and myelodysplasia in a patient with a complex chromosomal rearrangement involving the critical region 21q22. 2011 , 155A, 1697-705	14
271	Challenges in studying genomic structural variant formation mechanisms: the short-read dilemma and beyond. 2011 , 33, 840-50	30
270	Trisomic and allelic differences influence phenotypic variability during development of Down syndrome mice. 2011 , 189, 1487-95	15
269	Analysis of genomic variation in non-coding elements using population-scale sequencing data from the 1000 Genomes Project. 2011 , 39, 7058-76	58
268	Hirschsprung's disease in the neurologically challenged child. 2011 , 23, 223-7	3

267	syndrome phenotypes. 2011 , 33, 451-67	34
266	Down syndrome: searching for the genetic culprits. 2011 , 4, 586-95	74
265	The chromatin-binding protein HMGN1 regulates the expression of methyl CpG-binding protein 2 (MECP2) and affects the behavior of mice. 2011 , 286, 42051-42062	34
264	Over-expression of DSCAM and COL6A2 cooperatively generates congenital heart defects. 2011 , 7, e1002344	53
263	Genome-wide copy number analysis uncovers a new HSCR gene: NRG3. 2012 , 8, e1002687	46
262	Execretases, Alzheimer's Disease, and Down Syndrome. 2012 , 2012, 362839	21
261	Transcriptional consequences of aneuploidy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 12644-9	192
2 60	Genetic analysis of Down syndrome facilitated by mouse chromosome engineering. 2012 , 3, 8-12	8
259	Trisomy 21-associated defects in human primitive hematopoiesis revealed through induced pluripotent stem cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 17573-8	88
258	Pan-neuronal expression of APL-1, an APP-related protein, disrupts olfactory, gustatory, and touch plasticity in Caenorhabditis elegans. 2012 , 32, 10156-69	19
257	SNaPshot Assay in Quantitative Detection of Allelic Nondisjunction in Down Syndrome. 2012 , 16, 1226-35	2
256	Genetic modifiers predisposing to congenital heart disease in the sensitized Down syndrome population. 2012 , 5, 301-8	48
255	Mouse models of Down syndrome as a tool to unravel the causes of mental disabilities. 2012 , 2012, 584071	115
254	The use of mouse models for understanding the biology of down syndrome and aging. 2012 , 2012, 717315	6
253	Very low rate of gene conversion in the yeast genome. 2012 , 29, 3817-26	18
252	Inflammatory bowel disease manifesting after surgical treatment for Hirschsprung disease. 2012 , 55, 272-7	22
251	Leukemias in patients with Down syndrome. 503-519	
250	Down syndrome: the brain in trisomic mode. 2012 , 13, 844-58	159

249	DELLY: structural variant discovery by integrated paired-end and split-read analysis. 2012 , 28, i333-i339	1069
248	Activity-dependent phosphorylation of dynamin 1 at serine 857. 2012 , 51, 6786-96	8
247	Synaptic pathology of Down syndrome. 2012 , 970, 451-68	24
246	Deletion of a single-copy DAAM1 gene in congenital heart defect: a case report. 2012 , 13, 63	12
245	Pathways to cognitive deficits in Down syndrome. 2012 , 197, 73-100	34
244	A patient with Down syndrome with a de novo derivative chromosome 21. 2012 , 507, 159-64	6
243	Rectal biopsy in children with Down syndrome and chronic constipation: Hirschsprung disease vs non-hirschsprung disease. 2012 , 15, 87-95	16
242	Genomic determinants in the phenotypic variability of Down syndrome. 2012 , 197, 15-28	19
241	Paired tumor and normal whole genome sequencing of metastatic olfactory neuroblastoma. 2012 , 7, e37029	29
240	The Genetics of Fetal and Neonatal Cardiovascular Disease. 2012 , 343-376	
239	Increased dosage of the chromosome 21 ortholog Dyrk1a promotes megakaryoblastic leukemia in a murine model of Down syndrome. 2012 , 122, 948-62	110
238	Trisomy for synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. 2012 , 21, 3156-72	76
237	Chromosomal instability and aneuploidy in cancer: from yeast to man. 2012 , 13, 515-27	155
236	Rare copy number variants in isolated sporadic and syndromic atrioventricular septal defects. 2012 , 158A, 1279-84	35
235	No Evidence for Mutations that Deregulate GARS-AIRS-GART Protein Levels in Children with Down Syndrome. 2012 , 27, 46-51	1
234	Caenorhabditis elegans as a model organism to study APP function. 2012 , 217, 397-411	22
233	Spastic quadriplegia in Down syndrome with congenital duodenal stenosis/atresia. 2012, 52, 78-81	
232	Could submicroscopical chromosomal imbalances cause cono-truncal malformations in twins?. 2012 , 7, 170-7	1

231	Challenges to congenital genetic disorders with "RNA-targeting" chemical compounds. 2012, 134, 298-305	5
230	Amplified segment in the 'Down syndrome critical region' on HSA21 shared between Down syndrome and euploid AML-M0 excludes RUNX1, ERG and ETS2. 2012 , 157, 197-200	9
229	Etiopathological aspects of achalasia: lessons learned with Hirschsprung's disease. 2012, 25, 566-72	6
228	A rare de novo duplication of chromosome 21q22.12 -lq22.3 with other concomitant deletion and duplication of small fragments in 21q associated with Down syndrome: Prenatal diagnosis, molecular cytogenetic characterization. 2013 , 6, 11	4
227	Genome reassembly with high-throughput sequencing data. 2013 , 14 Suppl 1, S8	2
226	The amyloid precursor protein (APP) triplicated gene impairs neuronal precursor differentiation and neurite development through two different domains in the Ts65Dn mouse model for Down syndrome. 2013 , 288, 20817-20829	41
225	The impact of trisomy 21 on foetal haematopoiesis. 2013 , 51, 277-81	16
224	Enteric nervous system development: migration, differentiation, and disease. 2013, 305, G1-24	209
223	Surveying the Down syndrome mouse model resource identifies critical regions responsible for chronic otitis media. 2013 , 24, 439-45	7
222	Development of acute megakaryoblastic leukemia in Down syndrome is associated with sequential epigenetic changes. 2013 , 122, e33-43	36
221	Phenotypic impact of genomic structural variation: insights from and for human disease. 2013 , 14, 125-38	340
220	Child development and structural variation in the human genome. 2013 , 84, 34-48	17
219	Integration-free induced pluripotent stem cells model genetic and neural developmental features of down syndrome etiology. 2013 , 31, 467-78	107
218	Meeting at the crossroads: common mechanisms in Fragile X and Down syndrome. 2013 , 36, 685-94	12
217	Down Syndrome. 2013 , 414-417	
216	Global DNA hypermethylation in down syndrome placenta. 2013 , 9, e1003515	63
215	Down Syndrome. 2013 , 547-571	О
214	Trisomy 21 and facial developmental instability. 2013 , 151, 49-57	22

213	Aging and intellectual disability: insights from mouse models of Down syndrome. 2013 , 18, 43-50	22
212	Atypical aging in Down syndrome. 2013 , 18, 51-67	153
211	Concise review: new paradigms for Down syndrome research using induced pluripotent stem cells: tackling complex human genetic disease. 2013 , 2, 175-84	11
210	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. 2013 , 23, 1410-21	48
209	Chromosome 21 scan in Down syndrome reveals DSCAM as a predisposing locus in Hirschsprung disease. 2013 , 8, e62519	18
208	Echocardiography in children with Down syndrome. 2013 , 2, 36-45	12
207	Quantitative Evaluation of the Facial Morphology of a Tolteca Figurine from Mexico using Geometric Morphometric Approaches. 2014 , 32, 499-509	3
206	DYRK1A: a potential drug target for multiple Down syndrome neuropathologies. 2014 , 13, 26-33	77
205	Mapping genetically controlled neural circuits of social behavior and visuo-motor integration by a preliminary examination of atypical deletions with Williams syndrome. 2014 , 9, e104088	19
204	Opportunities and Limitations of Modelling Alzheimer's Disease with Induced Pluripotent Stem Cells. 2014 , 3, 1357-72	10
203	Identification of a DNA methylation signature in blood cells from persons with Down Syndrome. 2015 , 7, 82-96	68
202	Transcriptomics in Health and Disease. 2014 ,	О
201	Cognition and hippocampal plasticity in the mouse is altered by monosomy of a genomic region implicated in Down syndrome. 2014 , 197, 899-912	14
2 00	Tangram: a comprehensive toolbox for mobile element insertion detection. 2014 , 15, 795	39
199	The Molecular Genetics of Trisomy 18: PhenotypeLenotype Correlations. 2014 ,	3
198	Modelling and rescuing neurodevelopmental defect of Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. 2014 , 6, 259-77	117
197	Haematopoietic development and leukaemia in Down syndrome. 2014 , 167, 587-99	71
196	Engineered chromosome-based genetic mapping establishes a 3.7 Mb critical genomic region for Down syndrome-associated heart defects in mice. 2014 , 133, 743-53	22

(2015-2014)

195	A novel mouse model for Down syndrome that harbor a single copy of human artificial chromosome (HAC) carrying a limited number of genes from human chromosome 21. 2014 , 23, 317-29	12
194	Human chromosome 21 orthologous region on mouse chromosome 17 is a major determinant of Down syndrome-related developmental cognitive deficits. 2014 , 23, 578-89	27
193	Chromosomal syndromes. 2014 , 22, 197-203	2
192	Congenital heart disease protein 5 associates with CASZ1 to maintain myocardial tissue integrity. 2014 , 141, 3040-9	20
191	Morphometry of anatomical shape complexes with dense deformations and sparse parameters. 2014 , 101, 35-49	140
190	Aurea mediocritas: the importance of a balanced genome. 2014 , 6, a015842	16
189	Functional transcriptome analysis of the postnatal brain of the Ts1Cje mouse model for Down syndrome reveals global disruption of interferon-related molecular networks. 2014 , 15, 624	43
188	A patient with partial trisomy 21 and 7q deletion expresses mild Down syndrome phenotype. 2014 , 536, 441-3	18
187	Epigenetics: the neglected key to minimize learning and memory deficits in Down syndrome. 2014 , 45, 72-84	42
186	Pathological aggression. 86-96	
185	Down syndrome. 208-219	
184	Transcriptional Impact of Rare and Private Copy Number Variants in Hypoplastic Left Heart Syndrome. 2015 , 8, 682-9	11
183	"Down syndrome: an insight of the disease". 2015 , 22, 41	106
183 182	"Down syndrome: an insight of the disease". 2015 , 22, 41 Transient myeloproliferative disorder with partial trisomy 21. 2015 , 62, 2021-4	106 5
182	Transient myeloproliferative disorder with partial trisomy 21. 2015 , 62, 2021-4	5
182	Transient myeloproliferative disorder with partial trisomy 21. 2015 , 62, 2021-4 Partial trisomy 21: a fifty-year follow-up visit. 2015 , 167, 1610-3	5

177	Dissecting Alzheimer disease in Down syndrome using mouse models. 2015 , 9, 268	29
176	Inter-Dependent Mechanisms Behind Cognitive Dysfunction, Vascular Biology and Alzheimer's Dementia in Down Syndrome: Multi-Faceted Roles of APP. 2015 , 9, 299	3
175	Dosage of the Abcg1-U2af1 region modifies locomotor and cognitive deficits observed in the Tc1 mouse model of Down syndrome. 2015 , 10, e0115302	11
174	Protein dynamics associated with failed and rescued learning in the Ts65Dn mouse model of Down syndrome. 2015 , 10, e0119491	28
173	Cardio-STIC (spatio-temporal image correlation) as genetic ultrasound of fetal Down syndrome. 2015 , 28, 1943-9	4
172	Perturbations of heart development and function in cardiomyocytes from human embryonic stem cells with trisomy 21. 2015 , 33, 1434-46	19
171	Linking cell surface receptors to microtubules: tubulin folding cofactor D mediates Dscam functions during neuronal morphogenesis. 2015 , 35, 1979-90	25
170	Chronic up-regulation of the SHH pathway normalizes some developmental effects of trisomy in Ts65Dn mice. 2015 , 135, 68-80	14
169	A genetic cause of Alzheimer disease: mechanistic insights from Down syndrome. 2015 , 16, 564-74	301
168	Fish oil improves gene targets of Down syndrome in C57BL and BALB/c mice. 2015 , 35, 440-8	4
168 167	Fish oil improves gene targets of Down syndrome in C57BL and BALB/c mice. 2015 , 35, 440-8 A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015 , 563, 72-5	1
	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with	
167	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015 , 563, 72-5 Opposite phenotypes of muscle strength and locomotor function in mouse models of partial	1
167 166	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015 , 563, 72-5 Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. 2015 , 11, e1005062	23
167 166 165	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015, 563, 72-5 Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. 2015, 11, e1005062 Gene dosage imbalances: action, reaction, and models. 2015, 40, 309-17 Aneuploid proliferation defects in yeast are not driven by copy number changes of a few	1 23 42
167 166 165	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015, 563, 72-5 Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. 2015, 11, e1005062 Gene dosage imbalances: action, reaction, and models. 2015, 40, 309-17 Aneuploid proliferation defects in yeast are not driven by copy number changes of a few dosage-sensitive genes. 2015, 29, 898-903 Rescue of the abnormal skeletal phenotype in Ts65Dn Down syndrome mice using genetic and	1 23 42 40
167 166 165 164 163	A very rare case of trisomy 4q32.3-4q35.2 and trisomy 21q11.2-21q22.11 in a patient with recombinant chromosomes 4 and 21. 2015, 563, 72-5 Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. 2015, 11, e1005062 Gene dosage imbalances: action, reaction, and models. 2015, 40, 309-17 Aneuploid proliferation defects in yeast are not driven by copy number changes of a few dosage-sensitive genes. 2015, 29, 898-903 Rescue of the abnormal skeletal phenotype in Ts65Dn Down syndrome mice using genetic and therapeutic modulation of trisomic Dyrk1a. 2015, 24, 5687-96 Signal persistence and amplification in cancer development and possible, related opportunities for	1 23 42 40 28

159	The importance of understanding individual differences in Down syndrome. 2016 , 5,	98
158	Genetic dissection of Down syndrome-associated congenital heart defects using a new mouse mapping panel. 2016 , 5,	48
157	DYRK1A, a Dosage-Sensitive Gene Involved in Neurodevelopmental Disorders, Is a Target for Drug Development in Down Syndrome. 2016 , 10, 104	84
156	Identification of altered pathways in Down syndrome-associated congenital heart defects using an individualized pathway aberrance score. 2016 , 15,	5
155	Placental transcriptomes in the common aneuploidies reveal critical regions on the trisomic chromosomes and genome-wide effects. 2016 , 36, 812-22	8
154	Systematic reanalysis of partial trisomy 21 cases with or without Down syndrome suggests a small region on 21q22.13 as critical to the phenotype. 2016 , 25, 2525-2538	40
153	Systematic Cellular Disease Models Reveal Synergistic Interaction of Trisomy 21 and GATA1 Mutations in Hematopoietic Abnormalities. 2016 , 15, 1228-41	56
152	Dementia in Down's syndrome. 2016 , 15, 622-36	125
151	Aneuploidy: a common and early evidence-based biomarker for carcinogens and reproductive toxicants. 2016 , 15, 97	14
150	Generation of improved human cerebral organoids from single copy DYRK1A knockout induced pluripotent stem cells in trisomy 21: hypothetical solutions for neurodevelopmental models and therapeutic alternatives in down syndrome. 2016 , 40, 1256-1270	3
149	Alzheimer-related protein APL-1 modulates lifespan through heterochronic gene regulation in Caenorhabditis elegans. 2016 , 15, 1051-1062	11
148	Mouse models of Down syndrome: gene content and consequences. 2016 , 27, 538-555	78
147	Robust Yet Fragile: Expression Noise, Protein Misfolding, and Gene Dosage in the Evolution of Genomes. 2016 , 50, 113-131	20
146	A 2.78-Mb duplication on chromosome 21q22.11 implicates candidate genes in the partial trisomy 21 phenotype. 2016 , 1,	6
145	Mouse-based genetic modeling and analysis of Down syndrome. 2016 , 120, 111-122	17
144	The HSA21 gene EURL/C21ORF91 controls neurogenesis within the cerebral cortex and is implicated in the pathogenesis of Down Syndrome. 2016 , 6, 29514	15
143	Internal tandem duplication of FLT3 deregulates proliferation and differentiation and confers resistance to the FLT3 inhibitor AC220 by Up-regulating RUNX1 expression in hematopoietic cells. 2016 , 103, 95-106	8
142	Etiology of Acute Leukemias in Children. 2016 ,	1

Origin of Leukemia in Children with Down Syndrome. **2016**, 109-131

140	Widespread cerebellar transcriptome changes in Ts65Dn Down syndrome mouse model after lifelong running. 2016 , 296, 35-46	13
139	Some assembly required: evolutionary and systems perspectives on the mammalian reproductive system. 2016 , 363, 267-278	4
138	Noninvasive Prenatal Screening of Fetal Aneuploidy without Massively Parallel Sequencing. 2017 , 63, 861-869	7
137	Hematological Disorders in Children. 2017,	2
136	Genetics and Genomics of Congenital Heart Disease. 2017 , 120, 923-940	197
135	Down syndrome and the complexity of genome dosage imbalance. 2017 , 18, 147-163	143
134	Down Syndrome, Partial Trisomy 21, and Absence of Alzheimer's Disease: The Role of APP. 2017 , 56, 459-470	83
133	Pulmonary Hypertension in Adult Congenital Heart Disease. 2017,	6
132	Rodent models in Down syndrome research: impact and future opportunities. 2017 , 10, 1165-1186	87
131	Targeting trisomic treatments: optimizing Dyrk1a inhibition to improve Down syndrome deficits. 2017 , 5, 451-465	22
130	Eisenmenger Syndrome in Patients with Down Syndrome. 2017 , 279-289	1
129	The genetics of congenital heart disease understanding and improving long-term outcomes in congenital heart disease: a review for the general cardiologist and primary care physician. 2017 , 29, 520-528	18
128	Opposite chromosome constitutions due to a familial translocation t(1;21)(q43;q22) in 2 cousins with development delay and congenital anomalies: A case report. 2017 , 96, e6521	2
127	Genotype-phenotype correlation for congenital heart disease in Down syndrome through analysis of partial trisomy 21 cases. 2017 , 109, 391-400	19
126	Down syndrome: from the age of characterization to the era of curative approach. 2017 , 60, 197-208	2
125	Integrated Transcriptome Map Highlights Structural and Functional Aspects of the Normal Human Heart. 2017 , 232, 759-770	14
124	Cardioskeletal Muscle Disease Associated With Chromosomal Disorders. 2017 , 331-344	

(2019-2017)

123	Down's Syndrome and Triple Negative Breast Cancer: A Rare Occurrence of Distinctive Clinical Relationship. 2017 , 18,	3
122	CLOVE: classification of genomic fusions into structural variation events. 2017 , 18, 346	4
121	Aging With Down Syndrome: The Dual Diagnosis: Alzheimer's Disease and Down Syndrome. 2018 , 33, 253-262	10
120	Heart Disease and Stroke Statistics-2018 Update: A Report From the American Heart Association. 2018 , 137, e67-e492	3848
119	Hirschsprung disease - integrating basic science and clinical medicine to improve outcomes. 2018 , 15, 152-167	115
118	Dysregulation of neurotrophin signaling in the pathogenesis of Alzheimer disease and of Alzheimer disease in Down syndrome. 2018 , 114, 52-61	36
117	Dysfunction of autophagy and endosomal-lysosomal pathways: Roles in pathogenesis of Down syndrome and Alzheimer's Disease. 2018 , 114, 40-51	94
116	Can Green Tea Polyphenols Improve Phenotypes Associated With Down Syndrome?. 2018, 439-454	1
115	Down syndrome with posterior cortical atrophy. 2018 , 2018,	1
114	Advances in understanding the association between Down syndrome and Hirschsprung disease (DS-HSCR). 2018 , 34, 1127-1137	8
113	A prenatal case of partial trisomy 21 (q22.2q22.3), resulting from a paternal insertion translocation ins(16;21) and uncovered by QF-PCR, and characterized by array CGH and FISH. 2018 , 6, 1313-1316	O
112	The Impact of APP on Alzheimer-like Pathogenesis and Gene Expression in Down Syndrome iPSC-Derived Neurons. 2018 , 11, 32-42	38
111	Genetics and genomics of Down syndrome. 2019 , 1-39	5
110	Fetal Cardiovascular Disease. 2019 , 252-282	
109	Alzheimer Disease Pathogenesis: Insights From Molecular and Cellular Biology Studies of Oligomeric Aland Tau Species. 2019 , 13, 659	122
108	Heart Disease and Stroke Statistics-2019 Update: A Report From the American Heart Association. 2019 , 139, e56-e528	3937
107	Detection of 21q11.2-q22.11 deletions in a fetus by NIPT. 2019 , 33, e22711	3
106	Partial trisomy 21 map: Ten cases further supporting the highly restricted Down syndrome critical region (HR-DSCR) on human chromosome 21. 2019 , 7, e797	16

105	Congenital Anomalies and Genetic Associations in Hirschsprung Disease. 2019, 175-199	1
104	Exploring the Pathogenesis of Alzheimer Disease in Basal Forebrain Cholinergic Neurons: Converging Insights From Alternative Hypotheses. 2019 , 13, 446	68
103	New approaches to studying early brain development in Down syndrome. 2019, 61, 867-879	18
102	Lysosomal Dysfunction in Down Syndrome Is APP-Dependent and Mediated by APP-LITF (C99). 2019 , 39, 5255-5268	65
101	Down Syndrome. 2019 , 137-147	
100	DYRK1A regulates the recruitment of 53BP1 to the sites of DNA damage in part through interaction with RNF169. 2019 , 18, 531-551	16
99	Down syndrome. 2019 , 167, 321-336	14
98	Triple play of DYRK1A kinase in cortical progenitor cells of Trisomy 21. 2019 , 138, 19-25	6
97	Cbs overdosage is necessary and sufficient to induce cognitive phenotypes in mouse models of Down syndrome and interacts genetically with Dyrk1a. 2019 , 28, 1561-1577	21
96	DYRK1A and cognition: A lifelong relationship. 2019 , 194, 199-221	47
95	Bilingualism in children with a dual diagnosis of Down syndrome and Autism Spectrum Disorder. 2021 , 35, 663-689	0
94	Proteomics Study of Peripheral Blood Mononuclear Cells in Down Syndrome Children. 2020 , 9,	1
93	A multi-level developmental approach to exploring individual differences in Down syndrome: genes, brain, behaviour, and environment. 2020 , 104, 103638	8
92	Targeting Amyloidogenic Processing of APP in Alzheimer's Disease. 2020 , 13, 137	24
91	SON inhibits megakaryocytic differentiation via repressing RUNX1 and the megakaryocytic gene expression program in acute megakaryoblastic leukemia. 2021 , 28, 1000-1015	2
90	Gain of chromosome 21 in hematological malignancies: lessons from studying leukemia in children with Down syndrome. 2020 , 34, 1984-1999	11
89	Complex chromosomal rearrangements of human chromosome 21 in a patient manifesting clinical features partially overlapped with that of Down syndrome. 2020 , 139, 1555-1563	2
88		

(2021-2020)

87	Genetic and epigenetic pathways in Down syndrome: Insights to the brain and immune system from humans and mouse models. 2020 , 251, 1-28	9
86	Chromatin accessibility promotes hematopoietic and leukemia stem cell activity. 2020 , 11, 1406	15
85	Treatment of Epilepsy Associated with Common Chromosomal Developmental Diseases. 2020 , 15, 21-29	
84	Modeling Down syndrome in cells: From stem cells to organoids. 2020 , 251, 55-90	10
83	Heart Disease and Stroke Statistics-2020 Update: A Report From the American Heart Association. 2020 , 141, e139-e596	2824
82	Substrate interaction inhibits Becretase production of amyloid-peptides. 2020 , 56, 2578-2581	2
81	A Single, Shared Triploidy in Three Species of Parasitic Nematodes. 2020 , 10, 225-233	5
80	All Creatures Great and Small: New Approaches for Understanding Down Syndrome Genetics. 2021 , 37, 444-459	5
79	Targeting increased levels of APP in Down syndrome: Posiphen-mediated reductions in APP and its products reverse endosomal phenotypes in the Ts65Dn mouse model. 2021 , 17, 271-292	14
78	Molecular Mechanisms of Distinct Diseases.	
77	Heart Disease and Stroke Statistics-2021 Update: A Report From the American Heart Association. 2021 , 143, e254-e743	1087
76	Possible roles for the hominoid-specific DSCR4 gene in human cells. 2021 , 96, 1-11	2
75	Consequences of aneuploidy in human fibroblasts with trisomy 21. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021 , 118,	9
74	Multi-influential genetic interactions alter behaviour and cognition through six main biological cascades in Down syndrome mouse models. 2021 , 30, 771-788	8
73	C21orf91 Regulates Oligodendroglial Precursor Cell Fate-A Switch in the Glial Lineage?. 2021 , 15, 653075	3
72	Maternal antibodies facilitate Amyloid-Œlearance by activating Fc-receptor-Syk-mediated phagocytosis. 2021 , 4, 329	2
	the Providence of the Control of the Annual Control of the Control	
71	Insulin resistance, oxidative stress and mitochondrial defects in Ts65dn mice brain: A harmful synergistic path in down syndrome. 2021 , 165, 152-170	16

69	Molecular Mechanisms of the Genetic Predisposition to Acute Megakaryoblastic Leukemia in Infants With Down Syndrome. 2021 , 11, 636633	4
68	Congenital Heart Disease and Surgical Outcome in Down Syndrome.	
67	Association Analysis of Variants of and With Hirschsprung Disease Susceptibility in Han Chinese and Functional Evaluation in Zebrafish. 2021 , 9, 641152	0
66	Progress and Challenges in Laboratory-Based Diagnostic and Screening Approaches for Aneuploidy Detection during Pregnancy. 2021 , 26, 425-440	
65	Profile of down syndromelssociated malignancies: Epidemiology, clinical features and therapeutic aspects. 2021 , 6, 63-72	1
64	DSCAM/PAK1 pathway suppression reverses neurogenesis deficits in iPSC-derived cerebral organoids from patients with Down syndrome. 2021 , 131,	18
63	Basal Forebrain Cholinergic Neurons: Linking Down Syndrome and Alzheimer's Disease. 2021 , 13, 703876	1
62	Elevated soluble amyloid beta protofibrils in Down syndrome and Alzheimer's disease. 2021 , 114, 103641	6
61	Astrocytes in Down Syndrome Across the Lifespan. 2021 , 15, 702685	2
60	Transcriptional and Post-Transcriptional Regulations of Amyloid-Precursor Protein (APP) mRNA. 2021 , 2,	O
59	Context Fear Conditioning in Down Syndrome Mouse Models: Effects of Trisomic Gene Content, Age, Sex and Genetic Background. 2021 , 12,	0
58	Genetics of Alzheimer's disease in adults with Down syndrome. 2022 , 193-208	
57	Cascading Genetic and Environmental Effects on Development: Implications for Intervention. 275-288	4
56	Influence of allelic differences in Down syndrome. 2020 , 251, 29-54	2
55	DYRK1A regulates the recruitment of 53BP1 to the sites of DNA damage in part through interaction with RNF169.	1
54	Down syndrome mouse models have an abnormal enteric nervous system. 2019 , 5,	5
53	A collagen VI-dependent pathogenic mechanism for Hirschsprung's disease. 2015 , 125, 4483-96	60
52	Hirschsprung's disease, Down syndrome, and missing heritability: too much collagen slows migration. 2015 , 125, 4323-6	9

(2015-2015)

51	Heart Defects in Down Syndrome. 2015 , 21, 3334-42	3
50	Down syndrome iPSC model: endothelial perspective on tumor development. 2020 , 11, 3387-3404	2
49	Modular transcriptional repertoire and MicroRNA target analyses characterize genomic dysregulation in the thymus of Down syndrome infants. 2016 , 7, 7497-533	15
48	Germinal and Somatic Trisomy 21 Mosaicism: How Common is it, What are the Implications for Individual Carriers and How Does it Come About?. 2010 , 11, 409-19	34
47	Down syndrome: A curative prospect?. 2020 , 7, 168-193	5
46	Partial trisomy and tetrasomy of chromosome 21 without Down Syndrome phenotype and short overview of genotype-phenotype correlation. A case report. 2014 , 158, 321-5	16
45	Autosomal Aneuploidy. 2013 , 113-137	
44	Chromosomal Anomalies Associated with Congenital Heart Disease. 2014 , 47-71	
43	Thymus Gene Coexpression Networks: A Comparative Study in Children with and Without Down Syndrome. 2014 , 123-136	
42	Assembly of Non-Unique Insertion Content Using Next-Generation Sequencing. 2014 , 21-39	O
41	Inherited Bone Marrow Failure Syndrome, TAM. 2017 , 145-170	
40	The hominoid-specific gene DSCR4 is involved in regulation of human leukocyte migration.	
39	Cbsoverdosage is necessary and sufficient to induce cognitive phenotypes in mouse models of Down syndrome and interacts genetically withDyrk1a.	О
38	Multi-influential genetic interactions alter behaviour and cognition through six main biological cascades in Down syndrome mouse models.	О
37	Mapping behavioral landscapes in Down syndrome animal models. 2020 , 251, 145-179	
36	The Role of HSA21 Encoded Mirna in Down Syndrome Pathophysiology:Opportunities in miRNA-Targeted Pharmacotherapy and Diagnosis of the Down Syndrome. 2020 , 27, 302-312	
35	Maternal antibodies facilitate Amyloid-lælearance by activating Fc-receptor-Syk-mediated phagocytosis.	
34	Screening key genes associated with congenital heart defects in Down syndrome based on differential expression network. 2015 , 8, 8385-93	5

33	Impact of increased APP gene dose in Down syndrome and the Dp16 mouse model. 2021,	3
32	Aberrant crosstalk between insulin signaling and mTOR in young Down syndrome individuals revealed by neuronal-derived extracellular vesicles. 2021 ,	3
31	Heart Disease and Stroke Statistics-2022 Update: A Report From the American Heart Association 2022 , CIR000000000001052	196
30	Lowering levels of reelin in entorhinal cortex layer II-neurons results in lowered levels of intracellular amyloid-[]	O
29	Support vector machine learning and diffusion-derived structural networks predict amyloid quantity and cognition in adults with Down's syndrome 2022 ,	
28	Genetic Background Influences Severity of Colonic Aganglionosis and Response to GDNF Enemas in the Mouse Model of Hirschsprung Disease. 2021 , 22,	O
27	Structural Characterization of the Highly Restricted Down Syndrome Critical Region on 21q22.13: New and Transcript Isoforms 2021 , 12, 770359	2
26	APP and DYRK1A regulate axonal and synaptic vesicle protein networks and mediate Alzheimer's pathology in trisomy 21 neurons 2022 ,	2
25	Overexpression screen of chromosome 21 genes reveals modulators of Sonic hedgehog signaling relevant to Down syndrome.	
24	Psychosocial Risk Factors for Alzheimer Disease in Patients with Down Syndrome and Their Association with Brain Changes: A Narrative Review.	
23	Rodent Modeling of Alzheimer's Disease in Down Syndrome: In vivo and ex vivo Approaches. 16,	0
22	Craniofacial dysmorphology in Down Syndrome is caused by increased dosage of Dyrk1a and at least three other genes.	
21	Down syndrome: a model for chromosome abnormalities. 2023 , 45-68	
20	Implications of trisomy 21 on congenital features and health aspects. 2022 , 13-40	Ο
19	Development of specific phenotypes and genetic consequences in Down syndrome. 2022 , 135-180	0
18	Gene-dosage imbalance due to trisomic HSA21 and genotypephenotype association in Down syndrome. 2022 , 93-134	Ο
17	Indvivo and indvitro models for research on Down syndrome. 2022 , 405-464	0
16	Sex Differences in Protein Expression and Their Perturbations in Amniotic Fluid Cells of Down Syndrome Fetuses.	O

CITATION REPORT

15	Recurring germline mosaicism in a family due to reversion of an inherited derivative chromosome 8 from an 8;21 translocation with interstitial telomeric sequences. jmedgenet-2022-108586	0
14	Neurological and neurodevelopmental manifestations in children and adolescents with Down syndrome. 2022 ,	О
13	Therapeutics for mitochondrial dysfunction-linked diseases in down syndrome. 2023, 68, 25-43	0
12	Partial trisomy 21 with or without highly restricted Down yndrome critical region (HR-DSCR): report of two new cases and reanalysis of the genotype phenotype association. 2022 , 15,	o
11	Partial trisomy 21 with or without highly restricted-Down syndrome critical region (HR- DSCR). Report of two new cases and reanalysis of the genotype-phenotype association.	0
10	Dscam1 overexpression impairs the function of the gut nervous system in Drosophila.	1
9	Consequences of chromosome gain: A new view on trisomy syndromes. 2022 , 109, 2126-2140	O
8	Shaking up the silence: consequences of HMGN1 antagonizing PRC2 in the Down syndrome brain. 2022 , 15,	O
7	Dysregulated systemic metabolism in a Down syndrome mouse model. 2023, 68, 101666	0
6	Heart Disease and Stroke Statistics 2023 Update: A Report From the American Heart Association.	9
5	Genetics and Molecular Basis of Congenital Heart Defects in Down Syndrome: Role of Extracellular Matrix Regulation. 2023 , 24, 2918	0
4	Overexpression screen of chromosome 21 genes reveals modulators of Sonic hedgehog signaling relevant to Down syndrome. 2023 , 16,	0
3	Moving beyond amyloid and tau to capture the biological heterogeneity of Alzheimer⊠ disease. 2023 ,	О
2	Lowering levels of reelin in entorhinal cortex layer II-neurons results in lowered levels of intracellular amyloid-[12023, 5,	O
1	Craniofacial dysmorphology in Down syndrome is caused by increased dosage of Dyrk1a and at least three other genes. 2023 , 150,	0