Koenraad Devriendt

List of Publications by Year
in descending order

191	7,148	66343	76900
papers	citations	42 h-index	74 g-index

197	197	197	11731
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	GATA3 haplo-insufficiency causes human HDR syndrome. Nature, 2000, 406, 419-422.	27.8	516
2	Constitutively activating mutation in WASP causes X-linked severe congenital neutropenia. Nature Genetics, 2001, 27, 313-317.	21.4	401
3	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
4	Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. Nature Genetics, 2007, 39, 957-959.	21.4	284
5	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	2.8	238
6	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
7	Detection of genomic copy number changes in patients with idiopathic mental retardation by high-resolution X-array-CGH: important role for increased gene dosage ofXLMRgenes. Human Mutation, 2007, 28, 1034-1042.	2.5	166
8	Prenatal and pre-implantation genetic diagnosis. Nature Reviews Genetics, 2016, 17, 643-656.	16.3	155
9	Rare Variants in NR2F2 Cause Congenital Heart Defects in Humans. American Journal of Human Genetics, 2014, 94, 574-585.	6.2	146
10	Human laterality disorders. European Journal of Medical Genetics, 2006, 49, 349-362.	1.3	134
11	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
12	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118
13	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. European Journal of Human Genetics, 2015, 23, 1286-1293.	2.8	108
14	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	6.2	97
15	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	2.5	92
16	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	1.3	91
17	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87
18	Diaphragmatic hernia in Denysâ€Ðrash syndrome. American Journal of Medical Genetics Part A, 1995, 57, 97-101.	2.4	84

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19	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
20	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. Human Genetics, 2001, 108, 299-303.	3.8	77
21	Hydatidiform mole and triploidy: the role of genomic imprinting in placental development. Human Reproduction Update, 2005, 11, 137-142.	10.8	76
22	Isolated supravalvular aortic stenosis: functional haploinsufficiency of the elastin gene as a result of nonsense-mediated decay. Human Genetics, 2000, 106, 577-588.	3.8	67
23	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
24	A prospective study of the clinical utility of prenatal chromosomal microarray analysis in fetuses with ultrasound abnormalities and an exploration of a framework for reporting unclassified variants and risk factors. Genetics in Medicine, 2014, 16, 469-476.	2.4	66
25	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
26	Facial dysmorphism is influenced by ethnic background of the patient and of the evaluator. Clinical Genetics, 2017, 92, 166-171.	2.0	63
27	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, 753-764.	6.2	62
28	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	2.4	58
29	Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	2.3	58
30	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
31	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. Genetics in Medicine, 2021, 23, 1137-1142.	2.4	58
32	Interstitial telomeric sequences at the junction site of a jumping translocation. Human Genetics, 1997, 99, 735-737.	3.8	57
33	Criteria for HNF1B analysis in patients with congenital abnormalities of kidney and urinary tract. Nephrology Dialysis Transplantation, 2015, 30, 835-842.	0.7	57
34	Clinical and molecular genetic features of congenital spinal muscular atrophy. Annals of Neurology, 1996, 40, 731-738.	5.3	54
35	Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332.	2.5	53
36	Velocardiofacial Syndrome Patients with a Heterozygous Chromosome 22q11 Deletion Have Giant Platelets. Pediatric Research, 1998, 44, 607-611.	2.3	52

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37	A recognisable behavioural phenotype associated with terminal deletions of the short arm of chromosome 8. American Journal of Medical Genetics Part A, 1997, 74, 515-520.	2.4	49
38	Multiple lipomas linked to an RB1 gene mutation in a large pedigree with low penetrance retinoblastoma. European Journal of Human Genetics, 2001, 9, 690-694.	2.8	49
39	The diagnostic value of next generation sequencing in familial nonsyndromic congenital heart defects. American Journal of Medical Genetics, Part A, 2015, 167, 1822-1829.	1.2	48
40	MEIS2 involvement in cardiac development, cleft palate, and intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 1142-1146.	1.2	48
41	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. Genetics in Medicine, 2017, 19, 306-313.	2.4	47
42	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
43	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
44	Clinical delineation of the PACS1â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
45	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	3.8	43
46	Metal mining and birth defects: a case-control study in Lubumbashi, Democratic Republic of the Congo. Lancet Planetary Health, The, 2020, 4, e158-e167.	11.4	42
47	Comprehensive genome-wide analysis of routine non-invasive test data allows cancer prediction: A single-center retrospective analysis of over 85,000 pregnancies. EClinicalMedicine, 2021, 35, 100856.	7.1	42
48	A novel fragile X syndrome mutation reveals a conserved role for the carboxyâ€terminus in <scp>FMRP</scp> localization and function. EMBO Molecular Medicine, 2015, 7, 423-437.	6.9	41
49	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. Human Reproduction, 2017, 32, 687-697.	0.9	40
50	Exome sequencing identifies rare variants in multiple genes in atrioventricular septal defect. Genetics in Medicine, 2016, 18, 189-198.	2.4	39
51	Deletion in chromosome region 22q11 in a child with CHARGE association. Clinical Genetics, 1998, 53, 408-410.	2.0	38
52	Congenital heart defects in a novel recurrent 22q11.2 deletion harboring the genes CRKL and MAPK1. American Journal of Medical Genetics, Part A, 2012, 158A, 574-580.	1.2	38
53	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
54	Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891.	2.4	37

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55	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331.	2.5	37
56	Cohen syndrome: the clinical symptoms and stigmata at a young age. Clinical Genetics, 1996, 49, 237-241.	2.0	35
57	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
58	Partial DiGeorge syndrome in two patients with a 10p rearrangement. Clinical Genetics, 1999, 55, 269-276.	2.0	34
59	Familial deletions of chromosome 22q11: The Leuven experience. , 1998, 80, 531-532.		33
60	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	2.8	33
61	Defective DNA Polymerase Î±-Primase Leads to X-Linked Intellectual Disability Associated with Severe Growth Retardation, Microcephaly, and Hypogonadism. American Journal of Human Genetics, 2019, 104, 957-967.	6.2	32
62	Homozygous loss-of-function mutation in ALMS1 causes the lethal disorder mitogenic cardiomyopathy in two siblings. European Journal of Medical Genetics, 2014, 57, 532-535.	1.3	31
63	Genetic profile of isolated congenital diaphragmatic hernia revealed by targeted nextâ€generation sequencing. Prenatal Diagnosis, 2018, 38, 654-663.	2.3	31
64	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
65	De Novo Variants in LMNB1 Cause Pronounced Syndromic Microcephaly and Disruption of Nuclear Envelope Integrity. American Journal of Human Genetics, 2020, 107, 753-762.	6.2	30
66	Noninvasive prenatal diagnosis by genome-wide haplotyping of cell-free plasma DNA. Genetics in Medicine, 2020, 22, 962-973.	2.4	29
67	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. Genome Medicine, 2012, 4, 73.	8.2	28
68	Platelet studies in autism spectrum disorder patients and first-degree relatives. Molecular Autism, 2015, 6, 57.	4.9	28
69	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
70	Prenatal diagnosis of partial trisomy 3p(3p23â†'pter) and monosomy 7q(7q36â†'qter) in a fetus with microcephaly alobar holoprosencephaly and cyclopia. Prenatal Diagnosis, 1999, 19, 986-989.	2.3	26
71	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. Frontiers in Genetics, 2018, 9, 150.	2.3	26
72	Phenotypic spectrum of Auâ€“Kline syndrome: a report of six new cases and review of the literature. European Journal of Human Genetics, 2018, 26, 1272-1281.	2.8	26

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73	Non-Syndromic Cleft Lip with or without Cleft Palate: Genome-Wide Association Study in Europeans Identifies a Suggestive Risk Locus at 16p12.1 and Supports SH3PXD2A as a Clefting Susceptibility Gene. Genes, 2019, 10, 1023.	2.4	26
74	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26
75	Novel FRMD7 Mutations and Genomic Rearrangement Expand the Molecular Pathogenesis of X-Linked Idiopathic Infantile Nystagmus. Investigative Ophthalmology and Visual Science, 2015, 56, 1701-1710.	3.3	25
76	Malformations of the middle and inner ear on CT imaging in 22q11 deletion syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2975-2983.	1.2	25
77	Pseudoautosomal Region 1 Length Polymorphism in the Human Population. PLoS Genetics, 2014, 10, e1004578.	3.5	24
78	Presenting symptoms in adults with the 22q11 deletion syndrome. European Journal of Medical Genetics, 2014, 57, 157-162.	1.3	24
79	Expanding the phenotypic spectrum of PORCN variants in two males with syndromic microphthalmia. European Journal of Human Genetics, 2015, 23, 551-554.	2.8	24
80	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128.	2.3	24
81	Chromosomal phenotypes and submicroscopic abnormalities. Human Genomics, 2004, 1, 126-33.	2.9	23
82	Cerebellar Hypoplasia in a Patient with Veloâ€Cardioâ€Facial Syndrome. Developmental Medicine and Child Neurology, 1996, 38, 949-953.	2.1	23
83	Growth pattern in Kabuki syndrome with a KMT2D mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3172-3179.	1.2	23
84	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	2.4	23
85	Diaphragmatic hernia as the first echographic sign in Apert syndrome. , 2000, 20, 404-406.		22
86	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the GPR98 Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
87	Genotypic and phenotypic variation in six patients with solitary median maxillary central incisor syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2451-2458.	1.2	21
88	SPG20 mutation in three siblings with familial hereditary spastic paraplegia. Journal of Physical Education and Sports Management, 2017, 3, a001537.	1.2	21
89	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	2.0	21
90	Cliniciansâ€™ attitude towards family planning and timing of diagnosis in autosomal dominant polycystic kidney disease. PLoS ONE, 2017, 12, e0185779.	2.5	21

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91	A microduplication of CBP in a patient with mental retardation and a congenital heart defect. American Journal of Medical Genetics, Part A, 2007, 143A, 2160-2164.	1.2	20
92	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. European Journal of Medical Genetics, 2016, 59, 436-443.	1.3	20
93	Maternal liver transplant: Another cause of discordant fetal sex determination using cellâ€free DNA. Prenatal Diagnosis, 2018, 38, 148-150.	2.3	20
94	Nonâ€invasive prenatal testing suggesting a maternal malignancy: What do we tell the prospective parents in Belgium?. Prenatal Diagnosis, 2021, 41, 1264-1272.	2.3	20
95	Neuroblastoma in a mother and congenital central hypoventilation in her daughter: Variable expression of the same genetic disorder?. , 2000, 90, 430-431.		18
96	Variability in expression of a familial 2.79â€‰Mb microdeletion in chromosome14q22.1â€“22.2. American Journal of Medical Genetics, Part A, 2012, 158A, 1381-1387.	1.2	18
97	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	1.3	18
98	First trimester cystic hygroma colli: Retrospective analysis in a tertiary center. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 231, 60-64.	1.1	18
99	Compound heterozygous loss-of-function mutations in KIF20A are associated with a novel lethal congenital cardiomyopathy in two siblings. PLoS Genetics, 2018, 14, e1007138.	3.5	18
100	Prenatal diagnosis of a terminal short arm deletion of chromosome 8 in a fetus with an atrioventricular septal defect. , 1998, 18, 65-67.		17
101	Polyhydramnios as a prenatal symptom of the DiGeorge/velo-cardio-facial syndrome. , 1998, 18, 68-72.		17
102	Isolated terminal limb reduction defects: Extending the clinical spectrum of Adamsâ€“Oliver syndrome and ARHGAP31 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1576-1579.	1.2	17
103	Exome sequencing identifies ZFPM2 as a cause of familial isolated congenital diaphragmatic hernia and possibly cardiovascular malformations. European Journal of Medical Genetics, 2014, 57, 247-252.	1.3	17
104	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
105	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	3.5	17
106	No evidence of locus heterogeneity in familial microcephaly with or without chorioretinopathy, lymphedema, or mental retardation syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 52.	2.7	16
107	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	2.5	16
108	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. Genetics in Medicine, 2019, 21, 2774-2780.	2.4	16

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109	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	2.8	16
110	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
111	Genetic locus on chromosome 6p for multicystic renal dysplasia, pelvi-ureteral junction stenosis, and vesicoureteral reflux. American Journal of Medical Genetics Part A, 1995, 59, 396-397.	2.4	15
112	3 generation pedigree with paternal transmission of the 22q11.2 deletion syndrome: Intrafamilial phenotypic variability. European Journal of Medical Genetics, 2015, 58, 244-248.	1.3	15
113	HUWE1 mutation explains phenotypic severity in a case of familial idiopathic intellectual disability. European Journal of Medical Genetics, 2013, 56, 379-382.	1.3	14
114	Focus group discussions on secondary variants and next-generation sequencing technologies. European Journal of Medical Genetics, 2015, 58, 249-257.	1.3	14
115	Williamsâ€“Beuren syndrome: pitfalls for diagnosis in limited resources setting. Clinical Case Reports (discontinued), 2016, 4, 294-297.	0.5	14
116	Bilateral renal tumors in an adult man with Smith-Magenis syndrome: The role of the FLCN gene. European Journal of Medical Genetics, 2016, 59, 499-501.	1.3	14
117	The prenatal diagnosis of spinal muscular atrophy. , 1998, 18, 607-610.		13
118	Clinically relevant discordances identified after tertiary reassessment of fetuses with isolated congenital diaphragmatic hernia. Prenatal Diagnosis, 2017, 37, 883-888.	2.3	13
119	ZNF462 and KLF12 are disrupted by a de novo translocation in a patient with syndromic intellectual disability and autism spectrum disorder. European Journal of Medical Genetics, 2018, 61, 376-383.	1.3	13
120	Progressive extrapyramidal disorder with primary hypogonadism and alopecia in sibs: A new syndrome?. American Journal of Medical Genetics Part A, 1996, 62, 54-57.	2.4	12
121	Congenital sternoclavicular dermoid sinus. International Journal of Pediatric Otorhinolaryngology, 2016, 81, 65-67.	1.0	12
122	DNA Methylation Profiling and Genomic Analysis in 20 Children with Short Stature Who Were Born Small for Gestational Age. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4730-e4741.	3.6	12
123	Enhanced MCP-1 Release in Early Autosomal Dominant Polycystic Kidney Disease. Kidney International Reports, 2021, 6, 1687-1698.	0.8	12
124	Xâ€linked severe mental retardation and a progressive neurological disorder in a Belgian family: clinical and genetic studies. Clinical Genetics, 1997, 52, 155-161.	2.0	11
125	A complex Xp11.22 deletion in a patient with syndromic autism: Exploration of FAM120C as a positional candidate gene for autism. American Journal of Medical Genetics, Part A, 2014, 164, 3035-3041.	1.2	11
126	Mild humoral immunodeficiency in a patient with Xâ€linked Kabuki syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 801-803.	1.2	11

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127	LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	2.0	11
128	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	3.2	11
129	What should we tell parents? Congenital diaphragmatic hernia. Prenatal Diagnosis, 2022, 42, 398-407.	2.3	11
130	Semilobar holoprosencephaly in a 46,XY female fetus. Prenatal Diagnosis, 2001, 21, 839-841.	2.3	10
131	Duplication of the TGFBR1 gene causes features of Loeys-Dietz syndrome. European Journal of Medical Genetics, 2010, 53, 408-410.	1.3	10
132	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. European Journal of Human Genetics, 2016, 24, 51-58.	2.8	10
133	Association between sickle cell anemia and alpha thalassemia reveals a high prevalence of the Î±^{3.7} triplication in congolese patients than in worldwide series. Journal of Clinical Laboratory Analysis, 2018, 32, .	2.1	10
134	Fetal sex determination in twin pregnancies using non-invasive prenatal testing. Npj Genomic Medicine, 2019, 4, 15.	3.8	10
135	Next-generation sequencing in prenatal setting: Some examples of unexpected variant association. European Journal of Medical Genetics, 2020, 63, 103875.	1.3	10
136	Phenotypic spectrum of the <scp>RBM10</scp>â€mediated intellectual disability and congenital malformation syndrome beyond classic <scp>TARP</scp> syndrome features. Clinical Genetics, 2021, 99, 449-456.	2.0	10
137	Second trimester miscarriage of a male fetus with incontinentia pigmenti. , 1998, 80, 298-299.		9
138	X-linked adrenal hypoplasia congenita: a novel DAX1 missense mutation and challenges for clinical diagnosis in Africa. European Journal of Pediatrics, 2012, 171, 267-270.	2.7	9
139	Repeat genetic testing with targeted capture sequencing in primary arrhythmia syndrome and cardiomyopathy. European Journal of Human Genetics, 2017, 25, 1313-1323.	2.8	9
140	A novel FBXO28 frameshift mutation in a child with developmental delay, dysmorphic features, and intractable epilepsy: A second gene that may contribute to the 1q41â€q42 deletion phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1549-1558.	1.2	9
141	Large congenital follicular ovarian cyst in a girl with Kabuki syndrome. American Journal of Medical Genetics Part A, 1996, 65, 90-91.	2.4	8
142	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.7	8
143	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753.	0.9	8
144	ELEVATED MATERNAL SERUM AND AMNIOTIC FLUID ALPHA-FETOPROTEIN LEVELS IN THE DENYSâ€“DRASH SYNDROME. , 1996, 16, 455-457.		7

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145	Precocious puberty in Klinefelter syndrome: Non-specific result of neurological deficit?. , 1997, 72, 122-122.		7
146	A complex submicroscopic chromosomal imbalance in 19p13.11 with one microduplication and two microtriplications. European Journal of Medical Genetics, 2008, 51, 219-225.	1.3	7
147	Refining the locus of branchio-otic syndrome 2 (BOS2) to a 5.25ÂMb locus on chromosome 1q31.3q32.1. European Journal of Medical Genetics, 2009, 52, 393-397.	1.3	7
148	Congenital diaphragmatic hernia as a part of Nanceâ€“Horan syndrome?. European Journal of Human Genetics, 2018, 26, 359-366.	2.8	7
149	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909.	1.2	7
150	Holoprosencephaly: A case series from an area with high miningâ€related pollution. Birth Defects Research, 2019, 111, 1561-1563.	1.5	7
151	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	2.9	7
152	Regional localization of a gene for nonspecific XLMR to Xp11.3-p11.23 (MRX51) and tentative localization of an MRX gene to Xq23-q26.1. American Journal of Medical Genetics Part A, 1999, 85, 283-287.	2.4	6
153	Protective <scp>BCL</scp>11A and <scp>HBS</scp>1Lâ€<scp>MYB</scp> polymorphisms in a cohort of 102 Congolese patients suffering from sickle cell anemia. Journal of Clinical Laboratory Analysis, 2018, 32, .	2.1	6
154	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	3.1	6
155	Vestibular dysfunction is a manifestation of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 448-454.	1.2	6
156	Two cases of non-alcoholic fatty liver disease caused by biallelic ABHD5 mutations. Journal of Hepatology, 2020, 72, 1030-1032.	3.7	6
157	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	1.2	6
158	Two novel presentations of KCNMA1â€related pathologyâ€“â€“Expanding the clinical phenotype of a rare channelopathy. Molecular Genetics & Genomic Medicine, 2021, 9, e1797.	1.2	6
159	Heterogeneity in Omphalocoele With Absent Radial Ray Complex. , 1999, 82, 95-96.		5
160	Mirror-image gastroschisis in monochorionic female twins. European Journal of Medical Genetics, 2015, 58, 266-269.	1.3	5
161	Prenatally detected copy number variants in a national cohort: A postnatal followâ€up study. Prenatal Diagnosis, 2020, 40, 1272-1283.	2.3	5
162	Follow-up of an adult with Keutel syndrome. American Journal of Medical Genetics Part A, 1999, 85, 82-83.	2.4	4

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163	Velocardiofacial syndrome presenting as distal arthrogryposis. European Journal of Pediatrics, 2004, 163, 329-330.	2.7	4
164	Skinfold over toenail is pathognomonic for the popliteal pterygium syndrome in a Congolese family with large intrafamilial variability. Clinical Case Reports (discontinued), 2014, 2, 250-253.	0.5	4
165	A Novel Missense Variant in the PVRL4 Gene Underlying Ectodermal Dysplasia-Syndactyly Syndrome in a Turkish Child. Molecular Syndromology, 2018, 9, 22-24.	0.8	4
166	The sudden death of the combined first trimester aneuploidy screening, a single centre experience in Belgium. Clinical Chemistry and Laboratory Medicine, 2019, 57, e294-e297.	2.3	4
167	Genotype-phenotype correlations of UBA2 mutations in patients with ectrodactyly. European Journal of Medical Genetics, 2020, 63, 104009.	1.3	4
168	Phenotype and growth in Sotos syndrome patient from DR Congo (Central Africa). American Journal of Medical Genetics, Part A, 2020, 182, 1572-1575.	1.2	4
169	Homozygous missense STRADA mutation in a patient with polyhydramnios, megalencephaly and symptomatic epilepsy syndrome. Clinical Dysmorphology, 2021, 30, 121-124.	0.3	4
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