# 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 | What should we tell parents? Congenital diaphragmatic hernia. Prenatal Diagnosis, 2022, 42, 398-407. | 2.3 | 11 |
| 2 | DNA testing for sickle cell anemia in Africa: Implementation choices for the Democratic Republic of Congo. Journal of Clinical Laboratory Analysis, 2022, , e24398. | 2.1 | 2 |
| 3 | Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780. | 2.4 | 16 |
| 4 | 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 |
| 5 | Dysmorphism and major anomalies are a main predictor of survival in newborns admitted to the neonatal intensive care unit in the Democratic Republic of Congo. American Journal of Medical Genetics, Part A, 2021, 185, 453-460. | 1.2 | 1 |
| 6 | 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 |
| 7 | Dental and Craniofacial Characteristics in Patients With 14Q22.1-Q22.2 Deletion: A Case Series. Cleft Palate-Craniofacial Journal, 2021, 58, 505-513. | 0.9 | 1 |
| 8 | 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 |
| 9 | Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. Genetics in Medicine, 2021, 23, 1137-1142. | 2.4 | 58 |
| 10 | Homozygous missense STRADA mutation in a patient with polyhydramnios, megalencephaly and symptomatic epilepsy syndrome. Clinical Dysmorphology, 2021, 30, 121-124. | 0.3 | 4 |
| 11 | 8p21.3 deletions are rare causes of non-syndromic autism spectrum disorder. Neurogenetics, 2021, 22, 207-213. | 1.4 | 1 |
| 12 | Enhanced MCP-1 Release in Early Autosomal Dominant Polycystic Kidney Disease. Kidney International Reports, 2021, 6, 1687-1698. | 0.8 | 12 |
| 13 | 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 |
| 14 | Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679. | 3.5 | 17 |
| 15 | 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 |
| 16 | Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768. | 1.2 | 6 |
| 17 | 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 |
| 18 | Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510. | 7.9 | 87 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Extraocular muscle hypoplasia associated with Axenfeld-Rieger syndrome. Strabismus, 2021, 29, 216-220. | 0.7 | 0 |
| 20 | SEVERE CASE OF RENAL COLOBOMA SYNDROME IN LONG-TERM FOLLOW-UP. Retinal Cases and Brief Reports, 2020, 14, 77-81. | 0.6 | 2 |
| 21 | Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753. | 0.9 | 8 |
| 22 | Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464. | 2.5 | 17 |
| 23 | MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68. | 7.6 | 38 |
| 24 | Microdeletion of the entire IRF6 gene in a Subsaharian Africanâ€™s family with Van der Woude syndrome. Clinical Dysmorphology, 2020, 29, 24-27. | 0.3 | 2 |
| 25 | Agnathia otocephaly: A case from the Katanga Copperbelt. Birth Defects Research, 2020, 112, 1287-1291. | 1.5 | 1 |
| 26 | 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 |
| 27 | Genotype-phenotype correlations of UBA2 mutations in patients with ectrodactyly. European Journal of Medical Genetics, 2020, 63, 104009. | 1.3 | 4 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | Morphological characterization of newborns in Kinshasa, DR Congo: Common variants, minor, and major anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 632-639. | 1.2 | 1 |
| 32 | Two cases of non-alcoholic fatty liver disease caused by biallelic ABHD5 mutations. Journal of Hepatology, 2020, 72, 1030-1032. | 3.7 | 6 |
| 33 | Next-generation sequencing in prenatal setting: Some examples of unexpected variant association. European Journal of Medical Genetics, 2020, 63, 103875. | 1.3 | 10 |
| 34 | 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 |
| 35 | Noninvasive prenatal diagnosis by genome-wide haplotyping of cell-free plasma DNA. Genetics in Medicine, 2020, 22, 962-973. | 2.4 | 29 |
| 36 | LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600. | 2.0 | 11 |

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|---|---|---|---|
| 37 | The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355. | 3.2 | 11 |
| 38 | Prenatally detected copy number variants in a national cohort: A postnatal followâ€up study. Prenatal Diagnosis, 2020, 40, 1272-1283. | 2.3 | 5 |
| 39 | Usefulness of fragile X checklist and CGG distribution in specialized institutions in Kinshasa, DR Congo. Journal of Community Genetics, 2019, 10, 153-159. | 1.2 | 2 |
| 40 | Holoprosencephaly: A case series from an area with high miningâ€related pollution. Birth Defects Research, 2019, 111, 1561-1563. | 1.5 | 7 |
| 41 | Fetal sex determination in twin pregnancies using non-invasive prenatal testing. Npj Genomic Medicine, 2019, 4, 15. | 3.8 | 10 |
| 42 | PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814. | 2.4 | 58 |
| 43 | 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 |
| 44 | 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 |
| 45 | 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 |
| 46 | Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112. | 2.8 | 16 |
| 47 | Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3. | 3.1 | 6 |
| 48 | Congenital lateral abdominal wall defect in two Congolese children. Clinical Dysmorphology, 2019, 28, 50-52. | 0.3 | 0 |
| 49 | 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 |
| 50 | Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733. | 2.9 | 7 |
| 51 | Vestibular dysfunction is a manifestation of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 448-454. | 1.2 | 6 |
| 52 | Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563. | 4.5 | 118 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266. | 2.3 | 58 |
| 56 | 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 |
| 57 | Congenital diaphragmatic hernia as a part of Nanceâ€“Horan syndrome?. European Journal of Human Genetics, 2018, 26, 359-366. | 2.8 | 7 |
| 58 | Maternal liver transplant: Another cause of discordant fetal sex determination using cellâ€free DNA. Prenatal Diagnosis, 2018, 38, 148-150. | 2.3 | 20 |
| 59 | Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245. | 2.4 | 66 |
| 60 | Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975. | 2.4 | 67 |
| 61 | Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001. | 2.5 | 16 |
| 62 | 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 |
| 63 | Association between sickle cell anemia and alpha thalassemia reveals a high prevalence of the Î±<sup>3.7</sup> triplication in congolese patients than in worldwide series. Journal of Clinical Laboratory Analysis, 2018, 32, . | 2.1 | 10 |
| 64 | 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 |
| 65 | Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332. | 2.5 | 53 |
| 66 | 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 |
| 67 | Olfactory function in patients with nonsyndromic orofacial clefts and their unaffected relatives. American Journal of Medical Genetics, Part A, 2018, 176, 2375-2381. | 1.2 | 1 |
| 68 | The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128. | 2.3 | 24 |
| 69 | Genetic profile of isolated congenital diaphragmatic hernia revealed by targeted nextâ€generation sequencing. Prenatal Diagnosis, 2018, 38, 654-663. | 2.3 | 31 |
| 70 | 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 |
| 71 | 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 |
| 72 | 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 |

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|---|---|---|---|
| 73 | 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 |
| 74 | 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 |
| 75 | 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 |
| 76 | Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. Human Reproduction, 2017, 32, 687-697. | 0.9 | 40 |
| 77 | Clinically relevant discordances identified after tertiary reassessment of fetuses with isolated congenital diaphragmatic hernia. Prenatal Diagnosis, 2017, 37, 883-888. | 2.3 | 13 |
| 78 | Facial dysmorphism is influenced by ethnic background of the patient and of the evaluator. Clinical Genetics, 2017, 92, 166-171. | 2.0 | 63 |
| 79 | 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 |
| 80 | 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 |
| 81 | Wolf-Hirschhorn Syndrome: Clinical and Genetic Data from a First Case Diagnosed in Central Africa. Journal of Pediatric Genetics, 2017, 06, 186-190. | 0.7 | 1 |
| 82 | ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033. | 6.2 | 83 |
| 83 | SPG20 mutation in three siblings with familial hereditary spastic paraplegia. Journal of Physical Education and Sports Management, 2017, 3, a001537. | 1.2 | 21 |
| 84 | Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700. | 2.4 | 45 |
| 85 | 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 |
| 86 | Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331. | 2.5 | 37 |
| 87 | Cliniciansâ€™ attitude towards family planning and timing of diagnosis in autosomal dominant polycystic kidney disease. PLoS ONE, 2017, 12, e0185779. | 2.5 | 21 |
| 88 | Congenital sternoclavicular dermoid sinus. International Journal of Pediatric Otorhinolaryngology, 2016, 81, 65-67. | 1.0 | 12 |
| 89 | 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 |
| 90 | Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065. | 21.4 | 351 |

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|---|---|---|---|
| 91 | Williamsâ€“Beuren syndrome: pitfalls for diagnosis in limited resources setting. Clinical Case Reports (discontinued), 2016, 4, 294-297. | 0.5 | 14 |
| 92 | Growth pattern in Kabuki syndrome with a KMT2D mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3172-3179. | 1.2 | 23 |
| 93 | 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 |
| 94 | Prenatal and pre-implantation genetic diagnosis. Nature Reviews Genetics, 2016, 17, 643-656. | 16.3 | 155 |
| 95 | 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 |
| 96 | A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065. | 0.7 | 8 |
| 97 | Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663. | 3.3 | 35 |
| 98 | 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 |
| 99 | Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811. | 2.5 | 92 |
| 100 | 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 |
| 101 | Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891. | 2.4 | 37 |
| 102 | Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162. | 2.4 | 58 |
| 103 | Clinical delineation of the PACS1 â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675. | 1.2 | 44 |
| 104 | Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285. | 3.8 | 43 |
| 105 | Exome sequencing identifies rare variants in multiple genes in atrioventricular septal defect. Genetics in Medicine, 2016, 18, 189-198. | 2.4 | 39 |
| 106 | PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, . | 5.0 | 134 |
| 107 | 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 |
| 108 | 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 |

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| 109 | 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 |
| 110 | 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 |
| 111 | Focus group discussions on secondary variants and next-generation sequencing technologies. European Journal of Medical Genetics, 2015, 58, 249-257. | 1.3 | 14 |
| 112 | Criteria for HNF1B analysis in patients with congenital abnormalities of kidney and urinary tract. Nephrology Dialysis Transplantation, 2015, 30, 835-842. | 0.7 | 57 |
| 113 | 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 |
| 114 | Mirror-image gastroschisis in monochorionic female twins. European Journal of Medical Genetics, 2015, 58, 266-269. | 1.3 | 5 |
| 115 | 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 |
| 116 | 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 |
| 117 | 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 |
| 118 | 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 |
| 119 | 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 |
| 120 | Platelet studies in autism spectrum disorder patients and first-degree relatives. Molecular Autism, 2015, 6, 57. | 4.9 | 28 |
| 121 | MEIS2 involvement in cardiac development, cleft palate, and intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 1142-1146. | 1.2 | 48 |
| 122 | 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 |
| 123 | Pseudoautosomal Region 1 Length Polymorphism in the Human Population. PLoS Genetics, 2014, 10, e1004578. | 3.5 | 24 |
| 124 | Preaxial Polydactyly of the Foot: Variable Expression of Trisomy 13 in a Case from Central Africa. Case Reports in Genetics, 2014, 2014, 1-5. | 0.2 | 3 |
| 125 | Meningocele in a Congolese Female with Beckwith-Wiedemann Phenotype. Case Reports in Genetics, 2014, 2014, 1-4. | 0.2 | 2 |
| 126 | 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 |

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|---|---|---|---|
| 127 | 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 |
| 128 | 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 |
| 129 | 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 |
| 130 | A novel heterozygous mutation of three consecutive nucleotides causing Apert syndrome in a Congolese family. European Journal of Medical Genetics, 2014, 57, 169-173. | 1.3 | 3 |
| 131 | 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 |
| 132 | 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 |
| 133 | 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 |
| 134 | Rare Variants in NR2F2 Cause Congenital Heart Defects in Humans. American Journal of Human Genetics, 2014, 94, 574-585. | 6.2 | 146 |
| 135 | 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 |
| 136 | Presenting symptoms in adults with the 22q11 deletion syndrome. European Journal of Medical Genetics, 2014, 57, 157-162. | 1.3 | 24 |
| 137 | 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 |
| 138 | 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 |
| 139 | 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 |
| 140 | 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 |
| 141 | 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 |
| 142 | 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 |
| 143 | Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849. | 6.2 | 97 |
| 144 | Duplication of the TGFBR1 gene causes features of Loeys-Dietz syndrome. European Journal of Medical Genetics, 2010, 53, 408-410. | 1.3 | 10 |

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| 145 | 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 |
| 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 | 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 |
| 148 | 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 |
| 149 | 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 |
| 150 | Human laterality disorders. European Journal of Medical Genetics, 2006, 49, 349-362. | 1.3 | 134 |
| 151 | Gain-of-Function WASP Mutations in Pediatric and Adult Patients with Myelodysplasia or AML.. Blood, 2006, 108, 4516-4516. | 1.4 | 1 |
| 152 | Nephro-urologic, gastrointestinal, and ophthalmic findings. , 2005, , 105-122. | | 0 |
| 153 | Hydatidiform mole and triploidy: the role of genomic imprinting in placental development. Human Reproduction Update, 2005, 11, 137-142. | 10.8 | 76 |
| 154 | Early motor development in young children with 22q.11 deletion syndrome and a conotruncal heart defect. Developmental Medicine and Child Neurology, 2005, 47, 797-802. | 2.1 | 0 |
| 155 | Velocardiofacial syndrome presenting as distal arthrogryposis. European Journal of Pediatrics, 2004, 163, 329-330. | 2.7 | 4 |
| 156 | Chromosomal phenotypes and submicroscopic abnormalities. Human Genomics, 2004, 1, 126-33. | 2.9 | 23 |
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