# Christian R Marshall

List of Publications by Year
in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 135 papers | 14,996 citations | 44069 48 h-index | 20961 115 g-index |
| 144 all docs | 144 docs citations | 144 times ranked | 20265 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372. | 27.8 | 1,803 |
| 2 | Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488. | 6.2 | 1,641 |
| 3 | Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35. | 21.4 | 838 |
| 4 | Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694. | 6.2 | 819 |
| 5 | Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611. | 14.8 | 691 |
| 6 | Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214. | 21.4 | 641 |
| 7 | Contribution of SHANK3 Mutations to Autism Spectrum Disorder. American Journal of Human Genetics, 2007, 81, 1289-1297. | 6.2 | 604 |
| 8 | Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191. | 30.7 | 457 |
| 9 | Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. American Journal of Human Genetics, 2013, 93, 249-263. | 6.2 | 429 |
| 10 | Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443. | 2.4 | 404 |
| 11 | Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. JAMA - Journal of the American Medical Association, 2015, 314, 895. | 7.4 | 352 |
| 12 | Rare Copy Number Variation Discovery and Cross-Disorder Comparisons Identify Risk Genes for ADHD. Science Translational Medicine, 2011, 3, 95ra75. | 12.4 | 304 |
| 13 | Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, . | 3.8 | 295 |
| 14 | SHANK1 Deletions in Males with Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 879-887. | 6.2 | 292 |
| 15 | FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817. | 6.2 | 219 |
| 16 | Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710. | 3.8 | 200 |
| 17 | Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68. | 12.4 | 178 |
| 18 | A Discovery Resource of Rare Copy Number Variations in Individuals with Autism Spectrum Disorder. G3: Genes, Genomes, Genetics, 2012, 2, 1665-1685. | 1.8 | 175 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. American Journal of Human Genetics, 2018, 102, 142-155. | 6.2 | 156 |
| 20 | Copy number variations and risk for schizophrenia in 22q11.2 deletion syndrome. Human Molecular Genetics, 2008, 17, 4045-4053. | 2.9 | 155 |
| 21 | Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. PLoS Genetics, 2012, 8, e1002843. | 3.5 | 149 |
| 22 | Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. Nature Genetics, 2014, 46, 742-747. | 21.4 | 149 |
| 23 | Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768. | 2.9 | 140 |
| 24 | Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084. | 2.4 | 125 |
| 25 | Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Human Molecular Genetics, 2013, 22, 4485-4501. | 2.9 | 120 |
| 26 | Clinically relevant copy number variations detected in cerebral palsy. Nature Communications, 2015, 6, 7949. | 12.8 | 120 |
| 27 | A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26. | 3.8 | 118 |
| 28 | Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327. | 1.3 | 114 |
| 29 | Copy Number Variation in Obsessive-Compulsive Disorder and Tourette Syndrome: A Cross-Disorder Study. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 910-919. | 0.5 | 111 |
| 30 | Infantile Spasms Is Associated with Deletion of the MAGI2 Gene on Chromosome 7q11.23-q21.11. American Journal of Human Genetics, 2008, 83, 106-111. | 6.2 | 108 |
| 31 | Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157. | 2.4 | 103 |
| 32 | Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744. | 2.8 | 88 |
| 33 | Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510. | 7.9 | 87 |
| 34 | A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376. | 16.3 | 83 |
| 35 | De novo and rare inherited copy-number variations in the hemiplegic form of cerebral palsy. Genetics in Medicine, 2018, 20, 172-180. | 2.4 | 82 |
| 36 | Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063. | 7.2 | 77 |

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|---|---|---|---|
| 37 | A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752. | 2.4 | 73 |
| 38 | Detection and Characterization of Copy Number Variation in Autism Spectrum Disorder. Methods in Molecular Biology, 2012, 838, 115-135. | 0.9 | 72 |
| 39 | CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. Journal of Neurodevelopmental Disorders, 2014, 6, 9. | 3.1 | 71 |
| 40 | Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression. Genetics in Medicine, 2017, 19, 53-61. | 2.4 | 70 |
| 41 | 15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693. | 2.5 | 67 |
| 42 | Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. Journal of Pediatrics, 2016, 171, 313-316.e2. | 1.8 | 67 |
| 43 | Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47. | 3.8 | 67 |
| 44 | A microcosting and costâ€“consequence analysis of clinical genomic testing strategies in autism spectrum disorder. Genetics in Medicine, 2017, 19, 1268-1275. | 2.4 | 62 |
| 45 | PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517. | 7.6 | 58 |
| 46 | Copy Number Variable MicroRNAs in Schizophrenia and Their Neurodevelopmental Gene Targets. Biological Psychiatry, 2015, 77, 158-166. | 1.3 | 58 |
| 47 | Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot. Genetics in Medicine, 2019, 21, 1001-1007. | 2.4 | 58 |
| 48 | The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136. | 2.0 | 57 |
| 49 | Uncovering obsessive-compulsive disorder risk genes in a pediatric cohort by high-resolution analysis of copy number variation. Journal of Neurodevelopmental Disorders, 2016, 8, 36. | 3.1 | 55 |
| 50 | Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. G3: Genes, Genomes, Genetics, 2015, 5, 1775-1781. | 1.8 | 53 |
| 51 | The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in Medicine, 2020, 22, 1015-1024. | 2.4 | 51 |
| 52 | Mutations in RAB39B in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. Molecular Autism, 2017, 8, 59. | 4.9 | 49 |
| 53 | Complex Copy Number Variation of AMY1 does not Associate with Obesity in two East Asian Cohorts. Human Mutation, 2016, 37, 669-678. | 2.5 | 48 |
| 54 | CAOSâ€”Episodic Cerebellar Ataxia, Areflexia, Optic Atrophy, and Sensorineural Hearing Loss. Journal of Child Neurology, 2015, 30, 1749-1756. | 1.4 | 47 |

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|---|---|---|---|
| 55 | Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109. | 5.9 | 47 |
| 56 | Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathologica Communications, 2015, 3, 44. | 5.2 | 45 |
| 57 | 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 |
| 58 | Whole-Genome Sequencing Suggests Schizophrenia Risk Mechanisms in Humans with 22q11.2 Deletion Syndrome. G3: Genes, Genomes, Genetics, 2015, 5, 2453-2461. | 1.8 | 43 |
| 59 | Microdeletions ofELP4Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. Human Mutation, 2015, 36, 842-850. | 2.5 | 41 |
| 60 | Genome sequencing as a platform for pharmacogenetic genotyping: a pediatric cohort study. Npj Genomic Medicine, 2017, 2, 19. | 3.8 | 41 |
| 61 | Germline and somatic mutations in STXBP1 with diverse neurodevelopmental phenotypes. Neurology: Genetics, 2017, 3, e199. | 1.9 | 41 |
| 62 | OTX2mutations cause autosomal dominant pattern dystrophy of the retinal pigment epithelium. Journal of Medical Genetics, 2014, 51, 797-805. | 3.2 | 40 |
| 63 | Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406. | 1.2 | 40 |
| 64 | The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48. | 8.2 | 40 |
| 65 | Prospective cohort study for identification of underlying genetic causes in neonatal encephalopathy using whole-exome sequencing. Genetics in Medicine, 2018, 20, 486-494. | 2.4 | 38 |
| 66 | Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663. | 3.3 | 35 |
| 67 | ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148. | 1.9 | 35 |
| 68 | A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402. | 1.2 | 34 |
| 69 | Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. European Journal of Human Genetics, 2017, 25, 1303-1312. | 2.8 | 32 |
| 70 | Impact of DNA source on genetic variant detection from human whole-genome sequencing data. Journal of Medical Genetics, 2019, 56, 809-817. | 3.2 | 32 |
| 71 | De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026. | 2.4 | 32 |
| 72 | Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of MAGEL2-related disorders. Journal of Medical Genetics, 2018, 55, 316-321. | 3.2 | 31 |

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|---|---|---|---|
| 73 | Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. Fertility and Sterility, 2014, 101, 1079-1085.e3. | 1.0 | 30 |
| 74 | Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. European Journal of Human Genetics, 2015, 23, 310-316. | 2.8 | 30 |
| 75 | Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. American Journal of Human Genetics, 2016, 99, 1359-1367. | 6.2 | 30 |
| 76 | Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105. | 8.2 | 30 |
| 77 | Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880. | 2.6 | 28 |
| 78 | Bi-allelic mutations ofLONP1encoding the mitochondrial LonP1 protease cause pyruvate dehydrogenase deficiency and profound neurodegeneration with progressive cerebellar atrophy. Human Molecular Genetics, 2019, 28, 290-306. | 2.9 | 27 |
| 79 | Genome-wide rare copy number variations contribute to genetic risk for transposition of the great arteries. International Journal of Cardiology, 2016, 204, 115-121. | 1.7 | 26 |
| 80 | Estimated carrier frequency of creatine transporter deficiency in females in the general population using functional characterization of novel missense variants in the SLC6A8 gene. Gene, 2015, 565, 187-191. | 2.2 | 25 |
| 81 | Severe neurodegeneration, progressive cerebral volume loss and diffuse hypomyelination associated with a homozygous frameshift mutation in CSTB. European Journal of Human Genetics, 2017, 25, 775-778. | 2.8 | 24 |
| 82 | Carrier frequency of guanidinoacetate methyltransferase deficiency in the general population by functional characterization of missense variants in the GAMT gene. Molecular Genetics and Genomics, 2015, 290, 2163-2171. | 2.1 | 23 |
| 83 | Congenital myopathy with â€œcoronaâ€•fibres, selective muscle atrophy, and craniosynostosis associated with novel recessive mutations in SCN4A. Neuromuscular Disorders, 2017, 27, 574-580. | 0.6 | 23 |
| 84 | Genes and Pathways Implicated in Tetralogy of Fallot Revealed by Ultra-Rare Variant Burden Analysis in 231 Genome Sequences. Frontiers in Genetics, 2020, 11, 957. | 2.3 | 23 |
| 85 | Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847. | 6.2 | 22 |
| 86 | Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163. | 2.9 | 22 |
| 87 | MED23â€associated refractory epilepsy successfully treated with the ketogenic diet. American Journal of Medical Genetics, Part A, 2016, 170, 2421-2425. | 1.2 | 21 |
| 88 | Genome sequencing as a diagnostic test. Cmaj, 2021, 193, E1626-E1629. | 2.0 | 20 |
| 89 | Genome-wide tandem repeat expansions contribute to schizophrenia risk. Molecular Psychiatry, 2022, 27, 3692-3698. | 7.9 | 20 |
| 90 | Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736. | | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679. | 3.5 | 17 |
| 92 | Whole-genome sequencing suggests mechanisms for 22q11.2 deletion-associated Parkinsonâ€™s disease. PLoS ONE, 2017, 12, e0173944. | 2.5 | 17 |
| 93 | Complex genomic rearrangements in the dystrophin gene due to replicationâ€based mechanisms. Molecular Genetics & Genomic Medicine, 2014, 2, 539-547. | 1.2 | 16 |
| 94 | Microcephalyâ€capillary malformation syndrome: Brothers with a homozygous STAMBP mutation, uncovered by exome sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 3018-3022. | 1.2 | 16 |
| 95 | Use of clinical chromosomal microarray in Chinese patients with autism spectrum disorderâ€”implications of a copy number variation involving DPP10. Molecular Autism, 2017, 8, 31. | 4.9 | 16 |
| 96 | Genome sequencing broadens the range of contributing variants with clinical implications in schizophrenia. Translational Psychiatry, 2021, 11, 84. | 4.8 | 16 |
| 97 | De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730. | | 15 |
| 98 | Copy number variation in fetal alcohol spectrum disorder. Biochemistry and Cell Biology, 2018, 96, 161-166. | 2.0 | 15 |
| 99 | Clinical Genetic Risk Variants Inform a Functional Protein Interaction Network for Tetralogy of Fallot. Circulation Genomic and Precision Medicine, 2021, 14, e003410. | 3.6 | 15 |
| 100 | Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. Journal of Medical Genetics, 2022, 59, 571-578. | 3.2 | 14 |
| 101 | Recurrent duplications of the annexin A1 gene (ANXA1) in autism spectrum disorders. Molecular Autism, 2014, 5, 28. | 4.9 | 13 |
| 102 | Neuropsychiatric aspects of 22q11.2 deletion syndrome: considerations in the prenatal setting. Prenatal Diagnosis, 2017, 37, 61-69. | 2.3 | 13 |
| 103 | Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12. | 2.9 | 12 |
| 104 | Hereditary Mucin Deficiency Caused by Biallelic Loss of Function of MUC5B. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 761-768. | 5.6 | 12 |
| 105 | Diagnostic yield of genome sequencing for prenatal diagnosis of fetal structural anomalies. Prenatal Diagnosis, 2022, 42, 822-830. | 2.3 | 12 |
| 106 | Within-family influences on dimensional neurobehavioral traits in a high-risk genetic model. Psychological Medicine, 2022, 52, 3184-3192. | 4.5 | 11 |
| 107 | HLX is a candidate gene for a pattern of anomalies associated with congenital diaphragmatic hernia, short bowel, and asplenia. American Journal of Medical Genetics, Part A, 2017, 173, 3070-3074. | 1.2 | 10 |
| 108 | Genome sequencing for detection of pathogenic deep intronic variation: A clinical case report illustrating opportunities and challenges. American Journal of Medical Genetics, Part A, 2021, 185, 3129-3135. | 1.2 | 10 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91. | 3.8 | 9 |
| 110 | De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033. | 3.8 | 8 |
| 111 | Mild Idiopathic Infantile Hypercalcemiaâ€"Part 1: Biochemical and Genetic Findings. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 2915-2937. | 3.6 | 8 |
| 112 | Rare Copy Number Variants Identified Suggest the Regulating Pathways in Hypertension-Related Left Ventricular Hypertrophy. PLoS ONE, 2016, 11, e0148755. | 2.5 | 8 |
| 113 | A Chromosomal Duplication Encompassing Interleukin-33 Causes a Novel Hyper IgE Phenotype Characterized by Eosinophilic Esophagitis and Generalized Autoimmunity. Gastroenterology, 2022, 163, 510-513.e3. | 1.3 | 8 |
| 114 | Trio genome sequencing for developmental delay and pediatric heart conditions: A comparative microcost analysis. Genetics in Medicine, 2022, 24, 1027-1036. | 2.4 | 7 |
| 115 | Novel Population Specific Autosomal Copy Number Variation and Its Functional Analysis amongst Negritos from Peninsular Malaysia. PLoS ONE, 2014, 9, e100371. | 2.5 | 6 |
| 116 | Paternal uniparental disomy of chromosome 19 in a pair of monochorionic diamniotic twins with dysmorphic features and developmental delay. Journal of Medical Genetics, 2018, 55, 847-852. | 3.2 | 6 |
| 117 | Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3. | 3.1 | 6 |
| 118 | Genome sequencing among children with medical complexity: What constitutes value from parentsâ€™ perspective?. Journal of Genetic Counseling, 2022, 31, 523-533. | 1.6 | 5 |
| 119 | Arginine-Glycine Amidinotransferase Deficiency and Functional Characterization of Missense Variants inGATM. Human Mutation, 2016, 37, 926-932. | 2.5 | 4 |
| 120 | Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , . | 2.5 | 4 |
| 121 | Thiemann disease and familial digital arthropathy â€“ brachydactyly: two sides of the same coin?. Orphanet Journal of Rare Diseases, 2019, 14, 156. | 2.7 | 3 |
| 122 | Analysis of five deep-sequenced trio-genomes of the Peninsular Malaysia Orang Asli and North Borneo populations. BMC Genomics, 2019, 20, 842. | 2.8 | 3 |
| 123 | A novel intronic variant in UBE3A identified by genome sequencing in a patient with an atypical presentation of Angelman syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2145-2151. | 1.2 | 3 |
| 124 | Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748. | 2.0 | 3 |
| 125 | ISDN2014_0253: High resolution genomic analyses of a clinically defined autism spectrum disorder cohort. International Journal of Developmental Neuroscience, 2015, 47, 76-76. | 1.6 | 2 |
| 126 | MG-132â€…Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12. | 3.2 | 1 |

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|---|---|---|---|
| 127 | MG-129â€…Our experience ofin silicogene panel testing for clinically heterogeneous disorders using exome sequencing. Journal of Medical Genetics, 2015, 52, A11.1-A11. | 3.2 | 1 |
| 128 | Identification of a novel MSH6 germline variant in a family with multiple gastro-intestinal malignancies by next generation sequencing. Familial Cancer, 2015, 14, 69-75. | 1.9 | 1 |
| 129 | Genome sequencing identifies a rare case of moderate Zellweger spectrum disorder caused by a PEX3 defect: Case report and literature review. Molecular Genetics and Metabolism Reports, 2020, 25, 100664. | 1.1 | 1 |
| 130 | MG-108â€…Beyond the ACMG 56: Parental choices and initial results from a comprehensive whole genome sequencing-based search for predictive genomic variants in children. Journal of Medical Genetics, 2015, 52, A3.2-A4. | 3.2 | 0 |
| 131 | MG-130â€…Utilising whole exome sequencing to identify causative variants in genetically heterogeneous disorders. Journal of Medical Genetics, 2015, 52, A11.2-A11. | 3.2 | 0 |
| 132 | MG-106â€…Global developmental delay and characteristic facial features associated with pacs1 gene mutation â€“ report of two cases. Journal of Medical Genetics, 2015, 52, A1.2-A1. | 3.2 | 0 |
| 133 | MG-123â€…Exonic and intronic NRXN1 deletions: Novel genotype-phenotype correlations. Journal of Medical Genetics, 2015, 52, A9.1-A9. | 3.2 | 0 |
| 134 | MG-108â€…Agenesis of the corpus callosum and autism associated with zeb1 gene deletion â€“ a case report. Journal of Medical Genetics, 2015, 52, A2.1-A2. | 3.2 | 0 |
| 135 | Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i. | 1.2 | 0 |