# Marwan S Shinawi

List of Publications by Year
in descending order



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|---|---|---|---|
| 153 papers | 8,391 citations | 76326 40 h-index | 54911 84 g-index |
| 162 all docs | 162 docs citations | 162 times ranked | 14122 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454. | 2.9 | 7 |
| 2 | Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. Pediatric Neurology, 2022, 126, 65-73. | 2.1 | 8 |
| 3 | DNMT3Aovergrowth syndrome is associated with the development of hematopoietic malignancies in children and young adults. Blood, 2022, 139, 461-464. | 1.4 | 9 |
| 4 | Delineation of the 1q24.3 microdeletion syndrome provides further evidence for the potential role of non-coding RNAs in regulating the skeletal phenotype. Bone, 2021, 142, 115705. | 2.9 | 2 |
| 5 | De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660. | 2.4 | 20 |
| 6 | Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401. | 2.8 | 10 |
| 7 | Response to Mounts and Besser. Genetics in Medicine, 2021, 23, 240-242. | 2.4 | 1 |
| 8 | New Cohort of Patients With CEDNIK Syndrome Expands the Phenotypic and Genotypic Spectra. Neurology: Genetics, 2021, 7, e553. | 1.9 | 10 |
| 9 | Paroxysmal Kinesigenic Dyskinesia in Twins With Chromosome 16p11.2 Duplication Syndrome. Neurology: Genetics, 2021, 7, e549. | 1.9 | 1 |
| 10 | Biallelic <scp>ASCC1</scp> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (<scp>SMABF2</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 2190-2197. | 1.2 | 4 |
| 11 | Immunodeficiency and bone marrow failure with mosaic and germline TLR8 gain of function. Blood, 2021, 137, 2450-2462. | 1.4 | 47 |
| 12 | Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483. | 2.4 | 24 |
| 13 | Biallelic variants in RNU12 cause CDAGS syndrome. Human Mutation, 2021, 42, 1042-1052. | 2.5 | 5 |
| 14 | Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921. | 2.4 | 5 |
| 15 | Functional and epigenetic phenotypes of humans and mice with DNMT3A Overgrowth Syndrome. Nature Communications, 2021, 12, 4549. | 12.8 | 21 |
| 16 | Autosomal Dominant ANO5-Related Disorder Associated With Myopathy and Gnathodiaphyseal Dysplasia. Neurology: Genetics, 2021, 7, e612. | 1.9 | 4 |
| 17 | A phase 1/2 open label nonrandomized clinical trial of intravenous 2-hydroxypropyl-Î²-cyclodextrin for acute liver disease in infants with Niemann-Pick C1. Molecular Genetics and Metabolism Reports, 2021, 28, 100772. | 1.1 | 8 |
| 18 | Novel exon-skipping variant disrupting the basic domain of HCFC1 causes intellectual disability without metabolic abnormalities in both male and female patients. Journal of Human Genetics, 2021, 66, 717-724. | 2.3 | 10 |

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|---|---|---|---|
| 19 | Identification of disease-linked hyperactivating mutations in UBE3A through large-scale functional variant analysis. Nature Communications, 2021, 12, 6809. | 12.8 | 10 |
| 20 | Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. European Journal of Medical Genetics, 2020, 63, 103736. | 1.3 | 11 |
| 21 | Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. Clinical Genetics, 2020, 97, 437-446. | 2.0 | 10 |
| 22 | Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245. | 6.2 | 56 |
| 23 | Overcoming presynaptic effects of VAMP2 mutations with 4â€aminopyridine treatment. Human Mutation, 2020, 41, 1999-2011. | 2.5 | 11 |
| 24 | A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ¯/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077. | 6.2 | 23 |
| 25 | De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324. | 6.2 | 32 |
| 26 | Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226. | 2.4 | 22 |
| 27 | Abnormally increased carotid intima media-thickness and elasticity in patients with Morquio A disease. Orphanet Journal of Rare Diseases, 2020, 15, 73. | 2.7 | 5 |
| 28 | Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592. | 3.8 | 24 |
| 29 | Pathogenic variants in TNRC6B cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. Journal of Medical Genetics, 2020, 57, 717-724. | 3.2 | 14 |
| 30 | Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1133-1141. | 2.4 | 89 |
| 31 | AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094. | 12.8 | 150 |
| 32 | De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618. | 2.8 | 45 |
| 33 | CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733. | 2.4 | 48 |
| 34 | 2-Pyrrolidinone and Succinimide as Clinical Screening Biomarkers for GABA-Transaminase Deficiency: Anti-seizure Medications Impact Accurate Diagnosis. Frontiers in Neuroscience, 2019, 13, 394. | 2.8 | 23 |
| 35 | A mutation in Siteâ€1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. Molecular Genetics & Genomic Medicine, 2019, 7, e00733. | 1.2 | 13 |
| 36 | A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143. | 1.4 | 16 |

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|---|---|---|---|
| 37 | Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21. | 2.9 | 9 |
| 38 | Birth Defects Among 788 Children Born to Gulf War Veterans Based on Physical Examination. Journal of Occupational and Environmental Medicine, 2019, 61, 263-270. | 1.7 | 2 |
| 39 | Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234. | 2.8 | 15 |
| 40 | Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60. | 4.1 | 18 |
| 41 | Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924. | 6.2 | 23 |
| 42 | Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042. | 2.4 | 23 |
| 43 | De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552. | 6.2 | 19 |
| 44 | Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. Journal of Pediatric Hematology/Oncology, 2019, 41, 133-136. | 0.6 | 1 |
| 45 | Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363. | 2.9 | 11 |
| 46 | Inherited Deletion of 1q, Hyperparathyroidism and Signs of Y-chromosomal Influence in a Patient with Turner Syndrome. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 88-93. | 0.9 | 3 |
| 47 | WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43. | 6.2 | 88 |
| 48 | Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171. | 2.9 | 23 |
| 49 | Child Neurology: Brown-Vialetto-Van Laere syndrome. Neurology, 2018, 91, 938-941. | 1.1 | 10 |
| 50 | Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884. | 2.5 | 23 |
| 51 | DeSanto-Shinawi Syndrome: First Case in South America. Molecular Syndromology, 2018, 9, 154-158. | 0.8 | 11 |
| 52 | Mutations in the <scp>PH</scp> Domain of <scp>DNM</scp>1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. Molecular Genetics & Genomic Medicine, 2018, 6, 294-300. | 1.2 | 19 |
| 53 | Functional characterization of biallelic RTTN variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. Pediatric Research, 2018, 84, 435-441. | 2.3 | 11 |
| 54 | De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316. | 6.2 | 48 |

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|---|---|---|---|
| 55 | BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311. | 7.6 | 81 |
| 56 | De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013. | 7.6 | 67 |
| 57 | CpG Island Hypermethylation Mediated by DNMT3A Is a Consequence of AML Progression. Cell, 2017, 168, 801-816.e13. | 28.9 | 177 |
| 58 | Support for the Diagnosis of CHARGE Syndromeâ€"Reply. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 635. | 2.2 | 0 |
| 59 | The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048. | 2.4 | 85 |
| 60 | Adult-onset dystonia with marfanoid features. Neurology: Clinical Practice, 2017, 7, e31-e34. | 1.6 | 1 |
| 61 | CEDNIK. Child Neurology Open, 2017, 4, 2329048X1773321. | 1.1 | 16 |
| 62 | The spectrum of DNMT3A variants in Tattonâ€"Brownâ€"Rahman syndrome overlaps with that in hematologic malignancies. American Journal of Medical Genetics, Part A, 2017, 173, 3022-3028. | 1.2 | 42 |
| 63 | Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene ( GLDN ). Human Mutation, 2017, 38, 1477-1484. | 2.5 | 19 |
| 64 | Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371. | 2.5 | 27 |
| 65 | Prevalence of Semicircular Canal Hypoplasia in Patients With CHARGE Syndrome. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 168. | 2.2 | 24 |
| 66 | The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562. | 2.7 | 68 |
| 67 | Neuroligin 2 nonsense variant associated with anxiety, autism, intellectual disability, hyperphagia, and obesity. American Journal of Medical Genetics, Part A, 2017, 173, 213-216. | 1.2 | 68 |
| 68 | <scp>FBXL4</scp> defects are common in patients with congenital lactic acidemia and encephalomyopathic mitochondrial <scp>DNA</scp> depletion syndrome. Clinical Genetics, 2017, 91, 634-639. | 2.0 | 18 |
| 69 | Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 460-469. | 3.6 | 95 |
| 70 | Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593. | 1.2 | 21 |
| 71 | A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178. | 1.4 | 32 |
| 72 | TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. Nature Genetics, 2016, 48, 1359-1369. | 21.4 | 69 |

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|---|---|---|---|
| 73 | The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150. | 2.4 | 64 |
| 74 | Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357. | 6.2 | 98 |
| 75 | STXBP1 encephalopathy. Neurology, 2016, 86, 954-962. | 1.1 | 264 |
| 76 | Digynic triploidy: utility and challenges of noninvasive prenatal testing. Clinical Case Reports (discontinued), 2015, 3, 406-410. | 0.5 | 8 |
| 77 | Multiâ€systemic involvement in NGLY1â€related disorder caused by two novel mutations. American Journal of Medical Genetics, Part A, 2015, 167, 816-820. | 1.2 | 45 |
| 78 | WACloss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. Journal of Medical Genetics, 2015, 52, 754-761. | 3.2 | 41 |
| 79 | TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350. | 27.0 | 239 |
| 80 | Autosomal recessive posterior column ataxia with retinitis pigmentosa caused by novel mutations in theFLVCR1gene. International Journal of Neuroscience, 2015, 125, 43-49. | 1.6 | 21 |
| 81 | FGFR3-related condition: a skeletal dysplasia with similarities to thanatophoric dysplasia and SADDAN due to Lys650Met. Skeletal Radiology, 2015, 44, 441-445. | 2.0 | 5 |
| 82 | De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 1009. | 6.2 | 1 |
| 83 | A Novel Mutation in Isoform 3 of the Plasma Membrane Ca2+ Pump Impairs Cellular Ca2+ Homeostasis in a Patient with Cerebellar Ataxia and Laminin Subunit 1Î± Mutations. Journal of Biological Chemistry, 2015, 290, 16132-16141. | 3.4 | 41 |
| 84 | De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. American Journal of Human Genetics, 2015, 96, 682-690. | 6.2 | 48 |
| 85 | A 5-Month-Old Boy with Delay in Growth and Development and Decreased Muscle Tone. Clinical Chemistry, 2015, 61, 50-54. | 3.2 | 0 |
| 86 | Intragenic <scp>CAMTA1</scp> deletions are associated with a spectrum of neurobehavioral phenotypes. Clinical Genetics, 2015, 87, 478-482. | 2.0 | 18 |
| 87 | Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635. | 3.2 | 48 |
| 88 | Scoliosis and vertebral anomalies: Additional abnormal phenotypes associated with chromosome 16p11.2 rearrangement. American Journal of Medical Genetics, Part A, 2014, 164, 1118-1126. | 1.2 | 38 |
| 89 | Brain MRI abnormalities and spectrum of neurological and clinical findings in three patients with proximal 16p11.2 microduplication. American Journal of Medical Genetics, Part A, 2014, 164, 2003-2012. | 1.2 | 19 |
| 90 | Molecular and phenotypic characterization of atypical Williamsâ€“Beuren syndrome. Clinical Genetics, 2014, 86, 487-491. | 2.0 | 7 |

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|---|---|---|---|
| 91 | Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578. | 6.2 | 40 |
| 92 | Heterozygous 24-polyalanine repeats in the PHOX2B gene with different manifestations across three generations. Pediatric Pulmonology, 2014, 49, E13-E16. | 2.0 | 15 |
| 93 | Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. Gene, 2014, 537, 279-284. | 2.2 | 31 |
| 94 | NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay. American Journal of Medical Genetics, Part A, 2013, 161, 377-381. | 1.2 | 40 |
| 95 | Duplication of 20p12.3 associated with familial Wolffâ€“Parkinsonâ€“White syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 137-144. | 1.2 | 10 |
| 96 | Haploinsufficiency of ZNF238 is associated with corpus callosum abnormalities in 1q44 deletions. American Journal of Medical Genetics, Part A, 2013, 161, 711-716. | 1.2 | 28 |
| 97 | Transient Massive Trimethylaminuria Associated with Food Proteinâ€“Induced Enterocolitis Syndrome. JIMD Reports, 2013, 12, 11-15. | 1.5 | 13 |
| 98 | A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981. | 7.1 | 118 |
| 99 | Acute Intermittent Porphyria. Journal of Child Neurology, 2012, 27, 917-921. | 1.4 | 14 |
| 100 | Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2602-2605. | 1.2 | 13 |
| 101 | ADULT syndrome due to an R243W mutation in TP63. International Journal of Dermatology, 2012, 51, 693-696. | 1.0 | 12 |
| 102 | Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179. | 2.5 | 45 |
| 103 | Earlyâ€onset Hepatic Fibrosis in Lysinuric Protein Intolerance. Journal of Pediatric Gastroenterology and Nutrition, 2011, 53, 695-698. | 1.8 | 6 |
| 104 | Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia. European Journal of Human Genetics, 2011, 19, 152-156. | 2.8 | 47 |
| 105 | 11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. American Journal of Medical Genetics, Part A, 2011, 155, 1272-1280. | 1.2 | 84 |
| 106 | Desmosterolosisâ€”phenotypic and molecular characterization of a third case and review of the literature. American Journal of Medical Genetics, Part A, 2011, 155, 1597-1604. | 1.2 | 52 |
| 107 | Cobalamin F Disease Detected by Newborn Screening and Follow-up on a 14-Year-Old Patient. Pediatrics, 2011, 128, e1636-e1640. | 2.1 | 13 |
| 108 | Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988. | 2.9 | 74 |

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|---|---|---|---|
| 109 | Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. Genetics in Medicine, 2011, 13, 95-101. | 2.4 | 190 |
| 110 | Known and Possible Roles of Epigenetics in Autism. , 2011, , 737-755. | | 0 |
| 111 | McCune-Albright syndrome presenting with unilateral macroorchidism and bilateral testicular masses. Pediatric Radiology, 2010, 40, 16-20. | 2.0 | 6 |
| 112 | Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850. | 2.5 | 111 |
| 113 | Mixed gonadal dysgenesis in a child with isodicentric y chromosome: Does the relative proportion of the 45,X line really matter?. American Journal of Medical Genetics, Part A, 2010, 152A, 1832-1837. | 1.2 | 23 |
| 114 | Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341. | 3.2 | 447 |
| 115 | Increased Homocysteine in a Patient Diagnosed with Marfan Syndrome. Clinical Chemistry, 2010, 56, 1665-1668. | 3.2 | 3 |
| 116 | Progressive Myopathy With Multiple Symmetric Lipomatosis. Archives of Neurology, 2009, 66, 1576-7. | 4.5 | 2 |
| 117 | Mitochondrial Neurogastrointestinal Encephalopathy Due to Mutations in RRM2B. Archives of Neurology, 2009, 66, 1028-32. | 4.5 | 103 |
| 118 | Is this the Coffinâ€“Siris syndrome or the BOD syndrome?. American Journal of Medical Genetics, Part A, 2009, 149A, 559-562. | 1.2 | 12 |
| 119 | The Xp contiguous deletion syndrome and autism. American Journal of Medical Genetics, Part A, 2009, 149A, 1138-1148. | 1.2 | 19 |
| 120 | Atypical presentation of VLCAD deficiency associated with a novel ACADVL splicing mutation. Muscle and Nerve, 2009, 39, 374-382. | 2.2 | 23 |
| 121 | Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832. | 21.4 | 610 |
| 122 | A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271. | 21.4 | 171 |
| 123 | The MTHFR 677 Câ†'T polymorphism and behaviors in children with autism: exploratory genotypeâ€“phenotype correlations. Autism Research, 2009, 2, 98-108. | 3.8 | 57 |
| 124 | Microdeletions including YWHAE in the Miller-Dieker syndrome region on chromosome 17p13.3 result in facial dysmorphisms, growth restriction, and cognitive impairment. Journal of Medical Genetics, 2009, 46, 825-833. | 3.2 | 112 |
| 125 | Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders. Journal of Medical Genetics, 2009, 46, 382-388. | 3.2 | 213 |
| 126 | Lowâ€level mosaicism of trisomy 14: Phenotypic and molecular characterization. American Journal of Medical Genetics, Part A, 2008, 146A, 1395-1405. | 1.2 | 34 |

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|---|---|---|---|
| 127 | Delineation of the proximal 3q microdeletion syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1729-1735. | 1.2 | 15 |
| 128 | Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box small nucleolar RNA cluster. Nature Genetics, 2008, 40, 719-721. | 21.4 | 533 |
| 129 | Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471. | 21.4 | 535 |
| 130 | The array CGH and its clinical applications. Drug Discovery Today, 2008, 13, 760-770. | 6.4 | 171 |
| 131 | 15q13q14 deletions: Phenotypic characterization and molecular delineation by comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2008, 146A, 1933-1941. | 1.2 | 13 |
| 132 | Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. Blood, 2008, 112, 1042-1047. | 1.4 | 74 |
| 133 | Lymphedema of the Lower Extremity: Is It Genetic or Nongenetic?. Clinical Pediatrics, 2007, 46, 835-841. | 0.8 | 13 |
| 134 | Preaxial polydactyly in neurofibromatosis 1. Clinical Dysmorphology, 2007, 16, 193-194. | 0.3 | 6 |
| 135 | Hyperhomocysteinemia and cobalamin disorders. Molecular Genetics and Metabolism, 2007, 90, 113-121. | 1.1 | 14 |
| 136 | Multiple ganglion cysts (â€˜cystic ganglionosisâ€™): an unusual presentation in a child. Scandinavian Journal of Rheumatology, 2007, 36, 145-148. | 1.1 | 19 |
| 137 | A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). Genetics in Medicine, 2006, 8, 465-473. | 2.4 | 499 |
| 138 | Recognition of Smith-Lemli-Opitz syndrome (RSH) in the fetus: Utility of ultrasonography and biochemical analysis in pregnancies with low maternal serum estriol. American Journal of Medical Genetics, Part A, 2005, 138A, 56-60. | 1.2 | 19 |
| 139 | Extensive primary cutaneous herpes simplex virus type 1 infection in an infant following acute rotavirus gastroenteritis. European Journal of Pediatrics, 2005, 164, 175-176. | 2.7 | 0 |
| 140 | Autoantibodies against bactericidal/permeabilityâ€“increasing protein (BPIâ€“ANCA) in cystic fibrosis patients treated with azithromycin. Clinical and Experimental Medicine, 2005, 5, 80-85. | 3.6 | 20 |
| 141 | Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. Pediatric Pulmonology, 2005, 39, 374-378. | 2.0 | 19 |
| 142 | No live individual homozygous for a novel endoglin mutation was found in a consanguineous Arab family with hereditary haemorrhagic telangiectasia. Journal of Medical Genetics, 2004, 41, e119-e119. | 3.2 | 21 |
| 143 | Pulmonary manifestations and function tests in children genetically diagnosed with FMF. Pediatric Pulmonology, 2003, 35, 452-455. | 2.0 | 11 |
| 144 | The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. European Journal of Human Genetics, 2002, 10, 145-149. | 2.8 | 116 |

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|---|---|---|---|
| 145 | Familial mediterranean fever: The segregation of four different mutations in 13 individuals from one inbred family: Genotype-phenotype correlation and intrafamilial variability. American Journal of Medical Genetics Part A, 2002, 109, 198-201. | 2.4 | 15 |
| 146 | Crouzon syndrome: Association with absent pulmonary valve syndrome and severe tracheobronchomalacia. Pediatric Pulmonology, 2002, 34, 478-481. | 2.0 | 12 |
| 147 | The musculoskeletal manifestations of familial Mediterranean fever in children genetically diagnosed with the disease. Arthritis and Rheumatism, 2001, 44, 1416-1419. | 6.7 | 78 |
| 148 | Familial Mediterranean fever: prevalence, penetrance and genetic drift. European Journal of Human Genetics, 2001, 9, 634-637. | 2.8 | 146 |
| 149 | Familial Mediterranean fever: high gene frequency and heterogeneous disease among an Israeli-Arab population. Journal of Rheumatology, 2000, 27, 1492-5. | 2.0 | 23 |
| 150 | Familial Mediterranean Fever: Clinical and Genetic Characterization in a Mixed Pediatric Population of Jewish and Arab Patients. Pediatrics, 1999, 103, e70-e70. | 2.1 | 101 |
| 151 | Direct detection of common mutations in the familial Mediterranean fever gene (MEFV) using naturally occurring and primer mediated restriction fragment analysis. , 1999, 14, 91-91. | | 21 |
| 152 | CSF levels of carnitine in children with meningitis, neurologic disorders, acute gastroenteritis, and seizure. Neurology, 1998, 50, 1869-1871. | 1.1 | 21 |
| 153 | The differential contribution of MEFV mutant alleles to the clinical profile of familial Mediterranean fever. , 0, . | | 2 |