

# Andreas Tzschach

## List of Publications by Year in descending order

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134  
papers

7,974  
citations

66315

42  
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54882

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136  
all docs

136  
docs citations

136  
times ranked

13112  
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>PIGN</i> encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	2.6	4
2	Transitioning the Molecular Tumor Board from Proof of Concept to Clinical Routine: A German Single-Center Analysis. <i>Cancers</i> , 2021, 13, 1151.	1.7	27
3	The Importance of Extended Analysis Using Current Molecular Genetic Methods Based on the Example of a Cohort of 228 Patients with Hereditary Breast and Ovarian Cancer Syndrome. <i>Genes</i> , 2021, 12, 1483.	1.0	1
4	Marfan Syndrome Caused by Disruption of the <i>FBN1</i> Gene due to A Reciprocal Chromosome Translocation. <i>Genes</i> , 2021, 12, 1836.	1.0	11
5	Skeletal abnormalities are common features in <i>Aym</i> Gripp syndrome. <i>Clinical Genetics</i> , 2020, 97, 362-369.	1.0	10
6	New <i>Cav1.2</i> Channelopathy with High-Functioning Autism, Affective Disorder, Severe Dental Enamel Defects, a Short QT Interval, and a Novel <i>CACNA1C</i> Loss-of-Function Mutation. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8611.	1.8	10
7	Novel truncating <i>PPM1D</i> mutation in a patient with intellectual disability. <i>European Journal of Medical Genetics</i> , 2019, 62, 70-72.	0.7	10
8	<i>KCNC1</i> -related disorders: new de novo variants expand the phenotypic spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1319-1326.	1.7	43
9	<i>PUF60-SCRIB</i> fusion transcript in a patient with 8q24.3 microdeletion and atypical Verheij syndrome. <i>European Journal of Medical Genetics</i> , 2019, 62, 103587.	0.7	7
10	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in <i>ADNP</i> . <i>Biological Psychiatry</i> , 2019, 85, 287-297.	0.7	108
11	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	4.1	131
12	<i>Sema3a</i> plays a role in the pathogenesis of CHARGE syndrome. <i>Human Molecular Genetics</i> , 2018, 27, 1343-1352.	1.4	20
13	Variable clinical phenotype in two siblings with Aicardi-Goutières syndrome type 6 and a novel mutation in the <i>ADAR</i> gene. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 186-189.	0.7	18
14	Next-generation panel sequencing identifies <i>NF1</i> germline mutations in three patients with pheochromocytoma but no clinical diagnosis of neurofibromatosis type 1. <i>European Journal of Endocrinology</i> , 2018, 178, K1-K9.	1.9	19
15	Novel <i>VPS33B</i> mutation in a patient with autosomal recessive keratoderma ichthyosis deafness syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2862-2866.	0.7	8
16	Posterior amorphous corneal dystrophy in a patient with 12q21.33 deletion. <i>Ophthalmic Genetics</i> , 2018, 39, 645-647.	0.5	4
17	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of <i>MED12</i> -related disorders. <i>Clinical Genetics</i> , 2018, 94, 450-456.	1.0	24
18	Diagnostic value of partial exome sequencing in developmental disorders. <i>PLoS ONE</i> , 2018, 13, e0201041.	1.1	36

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19	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. <i>Human Mutation</i> , 2017, 38, 621-636.	1.1	54
20	Pierpont syndrome: report of a new patient. <i>Clinical Dysmorphology</i> , 2017, 26, 205-208.	0.1	12
21	Musculoskeletal Disease in MDA5-Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. <i>Arthritis and Rheumatology</i> , 2017, 69, 2081-2091.	2.9	44
22	Novel PRPS1 gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies. , 2017, 173, 2736-2742.		11
23	Skewed X-inactivation in a family with <i>DLG3</i> associated X-linked intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2545-2550.	0.7	12
24	Abstract 3391: Next generation sequencing paves the way for personalized medicine in pheochromocytoma and paraganglioma patients and their families. , 2017, , .		0
25	Novel ADAMTSL2-mutations in a patient with geleophysic dysplasia type I. <i>Clinical Dysmorphology</i> , 2016, 25, 106-109.	0.1	5
26	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. <i>Human Mutation</i> , 2016, 37, 847-864.	1.1	134
27	Interstitial 1q23.3q24.1 deletion in a patient with renal malformation, congenital heart disease, and mild intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2394-2399.	0.7	7
28	Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH. <i>Breast Cancer Research and Treatment</i> , 2016, 159, 585-590.	1.1	15
29	The molecular and phenotypic spectrum of <i>IQSEC2</i> -related epilepsy. <i>Epilepsia</i> , 2016, 57, 1858-1869.	2.6	46
30	Tentative clinical diagnosis of Lujan-Fryns syndrome "A conglomeration of different genetic entities?". <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 94-102.	0.7	11
31	Chromosome aberration associated with hippocampal impairment. <i>Psychiatry Research - Neuroimaging</i> , 2016, 254, 1-2.	0.9	0
32	Epilepsy is not a mandatory feature of STXBP1 associated ataxia-tremor-retardation syndrome. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 661-665.	0.7	30
33	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. <i>Molecular Psychiatry</i> , 2016, 21, 133-148.	4.1	243
34	<i>STXBP1</i> encephalopathy. <i>Neurology</i> , 2016, 86, 954-962.	1.5	264
35	BOD1 Is Required for Cognitive Function in Humans and Drosophila. <i>PLoS Genetics</i> , 2016, 12, e1006022.	1.5	18
36	Interstitial 1p32.1p32.3 deletion in a patient with multiple congenital anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2406-2410.	0.7	6

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37	Next-generation sequencing in X-linked intellectual disability. <i>European Journal of Human Genetics</i> , 2015, 23, 1513-1518.	1.4	112
38	Variants in <i>CUL4B</i> are Associated with Cerebral Malformations. <i>Human Mutation</i> , 2015, 36, 106-117.	1.1	37
39	Interstitial duplication of chromosome region 1q25.1q25.3: Report of a patient with mild cognitive deficits, tall stature and facial dysmorphisms. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 653-656.	0.7	4
40	Mutations in the histamine <i>N</i> -methyltransferase gene, <i>HNMT</i> , are associated with nonsyndromic autosomal recessive intellectual disability. <i>Human Molecular Genetics</i> , 2015, 24, 5697-5710.	1.4	27
41	Congenital CLN disease in two siblings. <i>Wiener Medizinische Wochenschrift</i> , 2015, 165, 210-213.	0.5	14
42	Abstract LB-044: Germline mutations in patients with hereditary breast and ovarian cancer establish <i>ERCC2</i> as a cancer susceptibility gene. , 2015, , .		0
43	Floating Harbor syndrome: <i>SRCAP</i> mutations are not restricted to exon 34. <i>Clinical Genetics</i> , 2014, 85, 498-499.	1.0	18
44	<i>De novo</i> partial deletion in <i>GRID2</i> presenting with complicated spastic paraplegia. <i>Muscle and Nerve</i> , 2014, 49, 289-292.	1.0	33
45	<i>PIK3R1</i> mutations in <i>SHORT</i> syndrome. <i>Clinical Genetics</i> , 2014, 86, 292-294.	1.0	31
46	Deletions in 14q24.1q24.3 are associated with congenital heart defects, brachydactyly, and mild intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 620-626.	0.7	16
47	Characterisation of <i>de novo</i> <i>MAPK10/JNK3</i> truncation mutations associated with cognitive disorders in two unrelated patients. <i>Human Genetics</i> , 2013, 132, 461-471.	1.8	48
48	Platelet defects in congenital variant of Rett syndrome patients with <i>FOXG1</i> mutations or reduced expression due to a position effect at 14q12. <i>European Journal of Human Genetics</i> , 2013, 21, 1349-1355.	1.4	13
49	Kohlschütter-Törz Syndrome: Mutations in <i>ROGDI</i> and Evidence of Genetic Heterogeneity. <i>Human Mutation</i> , 2013, 34, 296-300.	1.1	24
50	Clinical and functional characterisation of the combined respiratory chain defect in two sisters due to autosomal recessive mutations in <i>MTFMT</i> . <i>Mitochondrion</i> , 2013, 13, 743-748.	1.6	25
51	Novel <i>SLC9A6</i> mutations in two families with Christianson syndrome. <i>Clinical Genetics</i> , 2013, 83, 596-597.	1.0	24
52	Novel <i>WDR35</i> mutations in patients with cranioectodermal dysplasia (Sensenbrenner) Tj ETQq0 0 0 rgBT /Oyerlock 10 Tf 50 142	1.0	28
53	Xq22.3q23 deletion including <i>ACSL4</i> in a patient with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 860-864.	0.7	19
54	A novel <i>ALDH5A1</i> mutation is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1915-1922.	0.7	16

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55	Angelman syndrome and severe infections in a patient with de novo 15q11.2â€“q13.1 deletion and maternally inherited 2q21.3 microdeletion. <i>Gene</i> , 2013, 512, 453-455.	1.0	11
56	A mosaic maternal splice donor mutation in the EHMT1 gene leads to aberrant transcripts and to Kleeftstra syndrome in the offspring. <i>European Journal of Human Genetics</i> , 2013, 21, 887-890.	1.4	14
57	A Novel SLC6A8 Mutation in a Large Family with X-Linked Intellectual Disability: Clinical and Proton Magnetic Resonance Spectroscopy Data of Both Hemizygous Males and Heterozygous Females. <i>JIMD Reports</i> , 2013, 13, 91-99.	0.7	10
58	12q24.33 deletion: Report of a patient with intellectual disability and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1409-1413.	0.7	2
59	A newly recognized autosomal recessive syndrome affecting neurologic function and vision. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1207-1213.	0.7	9
60	X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 146.	1.2	22
61	Isolated NIBPL missense mutations that cause Cornelia de Lange syndrome alter MAU2 interaction. <i>European Journal of Human Genetics</i> , 2012, 20, 271-276.	1.4	24
62	Mirror-Image Asymmetry in Monozygotic Twins with Kabuki Syndrome. <i>Molecular Syndromology</i> , 2012, 3, 94-97.	0.3	9
63	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. <i>Lancet, The</i> , 2012, 380, 1674-1682.	6.3	940
64	Interstitial 3p25.3â€“p26.1 deletion in a patient with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2587-2590.	0.7	11
65	Parental Origin of de novo Cytogenetically Balanced Reciprocal Non-Robertsonian Translocations. <i>Cytogenetic and Genome Research</i> , 2012, 136, 242-245.	0.6	9
66	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 91, 694-702.	2.6	89
67	Interstitial 9q34.11â€“q34.13 deletion in a patient with severe intellectual disability, hydrocephalus, and cleft lip/palate. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1709-1712.	0.7	10
68	Mutations in NSUN2 Cause Autosomal- Recessive Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 90, 847-855.	2.6	243
69	Mutation of the conserved polyadenosine RNA binding protein, ZC3H14/dNab2, impairs neural function in <i>Drosophila</i> and humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 12390-12395.	3.3	77
70	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011, 478, 57-63.	13.7	805
71	Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. <i>European Journal of Human Genetics</i> , 2011, 19, 115-117.	1.4	52
72	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. <i>European Journal of Human Genetics</i> , 2011, 19, 717-720.	1.4	21

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73	Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2011, 89, 176-182.	2.6	73
74	ST3GAL3 Mutations Impair the Development of Higher Cognitive Functions. American Journal of Human Genetics, 2011, 89, 407-414.	2.6	89
75	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. Human Genetics, 2011, 129, 141-148.	1.8	45
76	Identification of a novel CDKL5 exon and pathogenic mutations in patients with severe mental retardation, early-onset seizures and Rett-like features. Neurogenetics, 2011, 12, 165-167.	0.7	34
77	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. BMC Medical Genetics, 2011, 12, 17.	2.1	25
78	A novel nonsense mutation in <i>TUSC3</i> is responsible for non-syndromic autosomal recessive mental retardation in a consanguineous Iranian family. American Journal of Medical Genetics, Part A, 2011, 155, 1976-1980.	0.7	43
79	Christianson syndrome in a patient with an interstitial Xq26.3 deletion. American Journal of Medical Genetics, Part A, 2011, 155, 2771-2774.	0.7	13
80	Novel <i>GDI1</i> mutation in a large family with nonsyndromic X-linked intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 3067-3070.	0.7	16
81	Cohen syndrome diagnosis using whole genome arrays. Journal of Medical Genetics, 2011, 48, 136-140.	1.5	29
82	A distinctive gene expression fingerprint in mentally retarded male patients reflects disease-causing defects in the histone demethylase KDM5C. PathoGenetics, 2010, 3, 2.	5.7	35
83	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. American Journal of Human Genetics, 2010, 86, 185-195.	2.6	220
84	Cranioectodermal Dysplasia, Sensenbrenner Syndrome, Is a Ciliopathy Caused by Mutations in the IFT122 Gene. American Journal of Human Genetics, 2010, 86, 949-956.	2.6	180
85	Brachyphalangy, polydactyly and tibial aplasia/hypoplasia syndrome (OMIM 609945): case report and review of the literature. European Journal of Pediatrics, 2010, 169, 1535-1539.	1.3	3
86	Reply to 5q35 duplication and Hunter-McAlpine syndrome: Missing the link. American Journal of Medical Genetics, Part A, 2010, 152A, 804-804.	0.7	0
87	Characterization of an interstitial 4q32 deletion in a patient with mental retardation and a complex chromosome rearrangement. American Journal of Medical Genetics, Part A, 2010, 152A, 1008-1012.	0.7	18
88	11q14.1-11q22.1 deletion in a 1-year-old male with minor dysmorphic features. American Journal of Medical Genetics, Part A, 2010, 152A, 2651-2655.	0.7	5
89	Chromosome aberrations involving 10q22: report of three overlapping interstitial deletions and a balanced translocation disrupting C10orf11. European Journal of Human Genetics, 2010, 18, 291-295.	1.4	22
90	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. European Journal of Human Genetics, 2010, 18, 539-543.	1.4	61

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91	Novel missense mutations in the ubiquitination-related gene <i>UBE2A</i> cause a recognizable X-linked mental retardation syndrome. <i>Clinical Genetics</i> , 2010, 77, 541-551.	1.0	45
92	A clinical and molecular genetic study of 112 Iranian families with primary microcephaly. <i>Journal of Medical Genetics</i> , 2010, 47, 823-828.	1.5	87
93	Interstitial deletion 2p11.2-p12: Report of a patient with mental retardation and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 242-245.	0.7	13
94	Autosomal dominant inheritance in a large family with focal facial dermal dysplasia (Brauer-Setleis) Tj ETQq0 0 0 rgBT /Overlock 10 TF	0.7	10
95	Craniosynostosis in a patient with 2q37.3 deletion 5q34 duplication: Association of extra copy of <i>MSX2</i> with craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1544-1549.	0.7	24
96	A balanced chromosomal translocation disrupting <i>ARHGEF9</i> is associated with epilepsy, anxiety, aggression, and mental retardation. <i>Human Mutation</i> , 2009, 30, 61-68.	1.1	131
97	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants of <i>COH1</i> . <i>Human Mutation</i> , 2009, 30, E404-E420.	1.1	44
98	Genetik der nichtsyndromalen geistigen Behinderung. <i>Medizinische Genetik</i> , 2009, 21, 231-236.	0.1	0
99	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. <i>The HUGO Journal</i> , 2009, 3, 41-49.	4.1	48
100	Hypergonadotropic hypogonadism in a patient with inv ins (2;4). <i>Journal of Developmental and Physical Disabilities</i> , 2009, 32, 226-230.	3.6	2
101	An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. <i>European Journal of Human Genetics</i> , 2009, 17, 125-128.	1.4	17
102	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 420-425.	1.4	79
103	Structural variation in Xq28: MECP2 duplications in 1% of patients with unexplained XLMR and in 2% of male patients with severe encephalopathy. <i>European Journal of Human Genetics</i> , 2009, 17, 444-453.	1.4	130
104	Identification of Mutations in TRAPPC9, which Encodes the NIK- and IKK- $\beta$ -Binding Protein, in Nonsyndromic Autosomal-Recessive Mental Retardation. <i>American Journal of Human Genetics</i> , 2009, 85, 909-915.	2.6	120
105	Characterization of interstitial Xp duplications in two families by tiling path array CGH. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 197-203.	0.7	35
106	Chromosome deletions in 13q33-p34: Report of four patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 337-342.	0.7	63
107	Czech dysplasia: Report of a large family and further delineation of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1859-1864.	0.7	18
108	MCT8 mutation analysis and identification of the first female with Allan-Herndon-Dudley syndrome due to loss of MCT8 expression. <i>European Journal of Human Genetics</i> , 2008, 16, 1029-1037.	1.4	56

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109	Identification of a nonsense mutation in the very low-density lipoprotein receptor gene (VLDLR) in an Iranian family with dysequilibrium syndrome. <i>European Journal of Human Genetics</i> , 2008, 16, 270-273.	1.4	50
110	Alopeciamental retardation syndrome: clinical and molecular characterization of four patients. <i>British Journal of Dermatology</i> , 2008, 159, ???-???.	1.4	4
111	A Defect in the TUSC3 Gene Is Associated with Autosomal Recessive Mental Retardation. <i>American Journal of Human Genetics</i> , 2008, 82, 1158-1164.	2.6	127
112	Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome in a Girl with Chromosome Translocation t(2;3)(q33;q23). <i>Ophthalmic Genetics</i> , 2008, 29, 37-40.	0.5	6
113	High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. <i>Journal of Medical Genetics</i> , 2008, 45, 704-709.	1.5	110
114	Mapping translocation breakpoints by next-generation sequencing. <i>Genome Research</i> , 2008, 18, 1143-1149.	2.4	118
115	A Defect in the Ionotropic Glutamate Receptor 6 Gene (GRIK2) Is Associated with Autosomal Recessive Mental Retardation. <i>American Journal of Human Genetics</i> , 2007, 81, 792-798.	2.6	137
116	Characterization of a 5.3 Mb deletion in 15q14 by comparative genomic hybridization using a whole genome tiling path BAC array in a girl with heart defect, cleft palate, and developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 172-178.	0.7	55
117	Radiation-Induced Late Effects in Two Affected Individuals of the Lilo Radiation Accident. <i>Radiation Research</i> , 2007, 167, 615-623.	0.7	29
118	Characterization of a 16 Mb interstitial chromosome 7q21 deletion by tiling path array CGH. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 333-337.	0.7	15
119	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. <i>Human Mutation</i> , 2007, 28, 207-208.	1.1	103
120	X-linked mental retardation: a comprehensive molecular screen of 47 candidate genes from a 7.4 Mb interval in Xp11. <i>European Journal of Human Genetics</i> , 2007, 15, 68-75.	1.4	19
121	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. <i>Human Genetics</i> , 2007, 121, 501-509.	1.8	116
122	Homozygosity mapping in consanguineous families reveals extreme heterogeneity of non-syndromic autosomal recessive mental retardation and identifies 8 novel gene loci. <i>Human Genetics</i> , 2007, 121, 43-48.	1.8	92
123	Heterotaxy and cardiac defect in a girl with chromosome translocation t(X;1)(q26;p13.1) and involvement of ZIC3. <i>European Journal of Human Genetics</i> , 2006, 14, 1317-1320.	1.4	16
124	A Novel MGST2 Non-Synonymous Mutation in a Chinese Pedigree with Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2006, 126, 1003-1005.	0.3	9
125	SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. <i>Human Genetics</i> , 2006, 118, 708-715.	1.8	67
126	A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oral-facial-digital type I syndrome. <i>Human Genetics</i> , 2006, 120, 171-178.	1.8	166



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127	NovelJARID1C/SMCX mutations in patients with X-linked mental retardation. Human Mutation, 2006, 27, 389-389.	1.1	120
128	Molecular cytogenetic analysis of a de novo interstitial deletion of 5q23.3q31.2 and its phenotypic consequences. American Journal of Medical Genetics, Part A, 2006, 140A, 496-502.	0.7	12
129	Molecular cytogenetic analysis of a de novo interstitial chromosome 10q22 deletion. American Journal of Medical Genetics, Part A, 2006, 140A, 1108-1110.	0.7	9
130	Molecular characterization of a balanced chromosome translocation in psoriasis vulgaris. Clinical Genetics, 2005, 69, 189-193.	1.0	8
131	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2005, 76, 227-236.	2.6	349
132	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	9.4	139
133	Molecular breakpoint analysis and relevance of variable mosaicism in a woman with short stature, primary amenorrhea, unilateral gonadoblastoma, and a 46,X,del(Y)(q11)/45,X karyotype. American Journal of Medical Genetics Part A, 2002, 112, 51-55.	2.4	3
134	ABSENCE OF Yq MICRODELETIONS IN INFERTILE MEN. Archives of Andrology, 2001, 47, 167-171.	1.0	11