Abdul Noor

List of Publications by Year
in descending order

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24 papers	5,758 citations	489802 18 h-index	799663 21 g-index
25 all docs	25 docs citations	25 times ranked	10607 citing authors

#	Article	IF	Citations
1	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. Journal of Medical Genetics, 2021, 58, 275-283.	1.5	14
2	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	1.6	35
3	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	1.1	67
4	MG-105â€...Delineating the phenotypes associated with the 15q11.2 BP1-BP2 deletion: Preliminary trends in psychometric evaluation. Journal of Medical Genetics, 2015, 52, A2.3-A3.	1.5	0
5	New recessive truncating mutation in LTBP3 in a family with oligodontia, short stature, and mitral valve prolapse. American Journal of Medical Genetics, Part A, 2015, 167, 1396-1399.	0.7	23
6	Current Tools for Interpretation of Genomic Copy Number Variants. Current Genetic Medicine Reports, 2015, 3, 202-208.	1.9	0
7	Copy number variant study of bipolar disorder in Canadian and UK populations implicates synaptic genes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 303-313.	1.1	76
8	Identification of risk genes for autism spectrum disorder through copy number variation analysis in Austrian families. Neurogenetics, 2014, 15, 117-127.	0.7	98
9	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.	1.4	140
10	Mutation in NSUN2, which Encodes an RNA Methyltransferase, Causes Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2012, 90, 856-863.	2.6	189
11	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	13.5	531
12	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
13	A novel deletion mutation in the TUSC3 gene in a consanguineous Pakistani family with autosomal recessive nonsyndromic intellectual disability. BMC Medical Genetics, 2011, 12, 56.	2.1	33
14	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
15	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
16	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	5.8	178
17	Characterization of a de novo translocation t(5;18)(q33.1;q12.1) in an autistic boy identifies a breakpoint close to SH3TC2, ADRB2, and HTR4 on 5q, and within the desmocollin gene cluster on 18q. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 817-826.	1.1	10
18	Oligodontia Is Caused by Mutation in LTBP3, the Gene Encoding Latent TGF-Î² Binding Protein 3. American Journal of Human Genetics, 2009, 84, 519-523.	2.6	79

#	Article	IF	Citations
19	Identification of Mutations in TRAPPC9, which Encodes the NIK- and IKK-Î²-Binding Protein, in Nonsyndromic Autosomal-Recessive Mental Retardation. American Journal of Human Genetics, 2009, 85, 909-915.	2.6	120
20	Copy number variation analysis and sequencing of the X-linked mental retardation gene TSPAN7/TM4SF2 in patients with autism spectrum disorder. Psychiatric Genetics, 2009, 19, 154-155.	0.6	19
21	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	2.6	1,641
22	CC2D2A, Encoding A Coiled-Coil and C2 Domain Protein, Causes Autosomal-Recessive Mental Retardation with Retinitis Pigmentosa. American Journal of Human Genetics, 2008, 82, 1011-1018.	2.6	88
23	CC2D2A, Encoding A Coiled-Coil and C2 Domain Protein, Causes Autosomal-Recessive Mental Retardation with Retinitis Pigmentosa. American Journal of Human Genetics, 2008, 83, 656.	2.6	2
24	Common Genetic Etiologies and Biological Pathways Shared Between Autism Spectrum Disorders and Intellectual Disabilities. , 0, , .		0