Hessa S Alsaif

List of Publications by Year
in descending order

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31 papers	886 citations	567281 15 h-index	501196 28 g-index
33 all docs	33 docs citations	33 times ranked	2027 citing authors

#	Article	IF	Citations
1	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
2	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
3	Two further cases of polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome, caused by a truncating variant in <scp>STRADA</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 604-607.	1.2	5
4	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
5	<scp>MYH1</scp> is a candidate gene for recurrent rhabdomyolysis in humans. American Journal of Medical Genetics, Part A, 2021, 185, 2131-2135.	1.2	8
6	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
7	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
8	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	3.8	1
9	Missense NAA20 variantsimpairing the NatB protein N-terminal acetyltransferase cause autosomal recessivedevelopmental delay, intellectual disability, and microcephaly. Genetics in Medicine, 2021, 23, 2213-2218.	2.4	11
10	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. American Journal of Human Genetics, 2021, 108, 1318-1329.	6.2	15
11	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
12	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161.	8.2	13
13	A de novo mutation in FMR1 in a patient with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103763.	1.3	4
14	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	1.2	7
15	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41
16	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. Nature Communications, 2020, 11, 2510.	12.8	31
17	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
18	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59

#	Article	IF	Citations
19	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. Human Genetics, 2020, 139, 513-519.	3.8	10
20	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
21	Homozygous Loss-of-Function Mutations in AP1B1, Encoding Beta-1 Subunit of Adaptor-Related Protein Complex 1, Cause MEDNIK-like Syndrome. American Journal of Human Genetics, 2019, 105, 1016-1022.	6.2	34
22	Homozygous lossâ€ofâ€function variants of TASP1 , a gene encoding an activator of the histone methyltransferases KMT2A and KMT2D, cause a syndrome of developmental delay, happy demeanor, distinctive facial features, and congenital anomalies. Human Mutation, 2019, 40, 1985-1992.	2.5	10
23	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
24	Identification of novel loci for pediatric cholestatic liver disease defined by KIF12, PPM1F, USP53, LSR, and WDR83OS pathogenic variants. Genetics in Medicine, 2019, 21, 1164-1172.	2.4	71
25	Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049.	3.8	29
26	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
27	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
28	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
29	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
30	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. American Journal of Human Genetics, 2017, 101, 603-608.	6.2	59
31	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	3.8	73