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	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
2	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
3	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	3.8	73
4	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
5	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
6	Female Infertility Caused by Mutations in the Oocyte-Specific Translational Repressor PATL2. American Journal of Human Genetics, 2017, 101, 603-608.	6.2	59
7	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59
8	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
9	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41
10	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
11	DALRD3 encodes a protein mutated in epileptic encephalopathy that targets arginine tRNAs for 3-methylcytosine modification. Nature Communications, 2020, 11, 2510.	12.8	31
12	Congenital glaucoma and CYP1B1: an old story revisited. Human Genetics, 2019, 138, 1043-1049.	3.8	29
13	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
14	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
15	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
16	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
17	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
18	Mutations in TP73 cause impaired mucociliary clearance and lissencephaly. American Journal of Human Genetics, 2021, 108, 1318-1329.	6.2	15

#	Article	IF	Citations
19	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. Genome Medicine, 2021, 13, 161.	8.2	13
20	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
21	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
22	Recessive mutations in SCYL2 cause a novel syndromic form of arthrogryposis in humans. Human Genetics, 2020, 139, 513-519.	3.8	10
23	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
24	<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
25	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
26	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	1.2	7
27	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
28	A de novo mutation in FMR1 in a patient with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103763.	1.3	4
29	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
30	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
31	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	3.8	1