Elisabeth Graf

List of Publications by Year
in descending order

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42 papers	4,483 citations	147801 31 h-index	254184 43 g-index
44 all docs	44 docs citations	44 times ranked	10687 citing authors

#	Article	IF	Citations
1	A de novo missense variant in GABRA4 alters receptor function in an epileptic and neurodevelopmental phenotype. Epilepsia, 2022, 63, .	5.1	6
2	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395.	2.9	7
3	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
4	De novo variants in neurodevelopmental disordersâ€"experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28.	2.0	64
5	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
6	Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. CRISPR Journal, 2021, 4, 178-190.	2.9	10
7	Identification and characterization of distinct brown adipocyte subtypes in C57BL/6J mice. Life Science Alliance, 2021, 4, e202000924.	2.8	14
8	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
9	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Reports, 2020, 31, 107647.	6.4	36
10	Mitochondrial Regulation of the 26S Proteasome. Cell Reports, 2020, 32, 108059.	6.4	28
11	Endogenous FGF21-signaling controls paradoxical obesity resistance of UCP1-deficient mice. Nature Communications, 2020, 11, 624.	12.8	60
12	Driver mutations in USP8 wild-type Cushingâ€™s disease. Neuro-Oncology, 2019, 21, 1273-1283.	1.2	65
13	Functional identity of hypothalamic melanocortin neurons depends on Tbx3. Nature Metabolism, 2019, 1, 222-235.	11.9	27
14	Mitochondrial DNA mutation analysis from exome sequencingâ€"A more holistic approach in diagnostics of suspected mitochondrial disease. Journal of Inherited Metabolic Disease, 2019, 42, 909-917.	3.6	57
15	Lessons from exome sequencing in prenatally diagnosed heart defects: A basis for prenatal testing. Clinical Genetics, 2019, 95, 582-589.	2.0	23
16	An atlas of the aging lung mapped by single cell transcriptomics and deep tissue proteomics. Nature Communications, 2019, 10, 963.	12.8	408
17	Intermuscular adipose tissue directly modulates skeletal muscle insulin sensitivity in humans. American Journal of Physiology - Endocrinology and Metabolism, 2019, 316, E866-E879.	3.5	97
18	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40

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19	Interplay of cellâ€“cell contacts and RhoA/ <scp>MRTF</scp> â€A signaling regulates cardiomyocyte identity. EMBO Journal, 2018, 37, .	7.8	66
20	Prediction of Adipose Browning Capacity by Systematic Integration of Transcriptional Profiles. Cell Reports, 2018, 23, 3112-3125.	6.4	57
21	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Reports, 2018, 44, 1-7.	1.5	15
22	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. European Journal of Human Genetics, 2017, 25, 960-965.	2.8	53
23	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
24	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	2.8	35
25	Molecular Integration of Incretin and Glucocorticoid Action Reverses Immunometabolic Dysfunction and Obesity. Cell Metabolism, 2017, 26, 620-632.e6.	16.2	66
26	Long-Term Cold Adaptation Does Not Require FGF21 or UCP1. Cell Metabolism, 2017, 26, 437-446.e5.	16.2	100
27	Exome sequencing reveals a novel homozygous splice site variant in the WNT1 gene underlying osteogenesis imperfecta type 3. Pediatric Research, 2017, 82, 753-758.	2.3	34
28	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
29	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
30	Systematic analysis of variants related to familial hypercholesterolemia in families with premature myocardial infarction. European Journal of Human Genetics, 2016, 24, 191-197.	2.8	70
31	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	6.2	77
32	Exome sequencing identifies a nonsense mutation in Fam46a associated with bone abnormalities in a new mouse model for skeletal dysplasia. Mammalian Genome, 2016, 27, 111-121.	2.2	27
33	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
34	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	6.2	86
35	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	6.2	110
36	Biallelic Mutations of Methionyl-tRNA Synthetase Cause a Specific Type of Pulmonary Alveolar Proteinosis Prevalent on RÃ©union Island. American Journal of Human Genetics, 2015, 96, 826-831.	6.2	94

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37	Haploinsufficiency of TBK1 causes familial ALS and fronto-temporal dementia. Nature Neuroscience, 2015, 18, 631-636.	14.8	652
38	Mutations in the deubiquitinase gene USP8 cause Cushing's disease. Nature Genetics, 2015, 47, 31-38.	21.4	450
39	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	6.2	55
40	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
41	Dysfunctional nitric oxide signalling increases risk of myocardial infarction. Nature, 2013, 504, 432-436.	27.8	230
42	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. American Journal of Human Genetics, 2011, 88, 788-795.	6.2	206