# Elisabeth Graf

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2930973/publications.pdf

Version: 2024-02-01

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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 | Haploinsufficiency of TBK1 causes familial ALS and fronto-temporal dementia. Nature Neuroscience, 2015, 18, 631-636. | 14.8 | 652 |
| 2 | Mutations in the deubiquitinase gene USP8 cause Cushing's disease. Nature Genetics, 2015, 47, 31-38. | 21.4 | 450 |
| 3 | Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824. | 12.8 | 432 |
| 4 | An atlas of the aging lung mapped by single cell transcriptomics and deep tissue proteomics. Nature Communications, 2019, 10, 963. | 12.8 | 408 |
| 5 | Dysfunctional nitric oxide signalling increases risk of myocardial infarction. Nature, 2013, 504, 432-436. | 27.8 | 230 |
| 6 | 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 |
| 7 | Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918. | 10.2 | 139 |
| 8 | 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 |
| 9 | 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 |
| 10 | Long-Term Cold Adaptation Does Not Require FGF21 or UCP1. Cell Metabolism, 2017, 26, 437-446.e5. | 16.2 | 100 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, . | 8.2 | 89 |
| 16 | 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 |
| 17 | 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 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Molecular Integration of Incretin and Glucocorticoid Action Reverses Immunometabolic Dysfunction and Obesity. Cell Metabolism, 2017, 26, 620-632.e6. | 16.2 | 66 |
| 20 | Interplay of cellâ€“cell contacts and RhoA/ <scp>MRTF</scp> â€A signaling regulates cardiomyocyte identity. EMBO Journal, 2018, 37, . | 7.8 | 66 |
| 21 | 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 |
| 22 | Driver mutations in USP8 wild-type Cushingâ€™s disease. Neuro-Oncology, 2019, 21, 1273-1283. | 1.2 | 65 |
| 23 | De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28. | 2.0 | 64 |
| 24 | Endogenous FGF21-signaling controls paradoxical obesity resistance of UCP1-deficient mice. Nature Communications, 2020, 11, 624. | 12.8 | 60 |
| 25 | Prediction of Adipose Browning Capacity by Systematic Integration of Transcriptional Profiles. Cell Reports, 2018, 23, 3112-3125. | 6.4 | 57 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | 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 |
| 32 | 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 |
| 33 | Mitochondrial Regulation of the 26S Proteasome. Cell Reports, 2020, 32, 108059. | 6.4 | 28 |
| 34 | 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 |
| 35 | Functional identity of hypothalamic melanocortin neurons depends on Tbx3. Nature Metabolism, 2019, 1, 222-235. | 11.9 | 27 |
| 36 | Lessons from exome sequencing in prenatally diagnosed heart defects: A basis for prenatal testing. Clinical Genetics, 2019, 95, 582-589. | 2.0 | 23 |

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|---|---|---|---|
| 37 | 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 |
| 38 | Identification and characterization of distinct brown adipocyte subtypes in C57BL/6J mice. Life Science Alliance, 2021, 4, e202000924. | 2.8 | 14 |
| 39 | Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. CRISPR Journal, 2021, 4, 178-190. | 2.9 | 10 |
| 40 | 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 |
| 41 | A de novo missense variant in GABRA4 alters receptor function in an epileptic and neurodevelopmental phenotype. Epilepsia, 2022, 63, . | 5.1 | 6 |
| 42 | Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395. | 2.4 | 4 |