Katrin Ounap

List of Publications by Year in descending order

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147 papers	6,899 citations	87843 38 h-index	79644 73 g-index
152 all docs	152 docs citations	152 times ranked	12053 citing authors

#	Article	IF	Citations
1	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. Nature, 2010, 463, 671-675.	13.7	476
2	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
3	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurology, The, 2011, 10, 806-818.	4.9	352
4	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
5	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	1.5	251
6	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	1.5	250
7	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
8	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	6.0	195
9	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.5	172
10	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
11	Epigenetic mutations of the imprinted IGF2-H19 domain in Silver-Russell syndrome (SRS): results from a large cohort of patients with SRS and SRS-like phenotypes. Journal of Medical Genetics, 2008, 46, 192-197.	1.5	138
12	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and followâ€up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176.	1.7	132
13	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. Journal of Medical Genetics, 2011, 48, 105-116.	1.5	123
14	Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. Journal of Medical Genetics, 2013, 50, 463-472.	1.5	122
15	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
16	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	2.6	111
17	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	1.4	108
18	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.5	97

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19	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	2.6	93
20	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	1.7	91
21	Genotypeâ€“phenotype correlations in patients with retinoblastoma and interstitial 13q deletions. European Journal of Human Genetics, 2011, 19, 947-958.	1.4	83
22	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. Human Molecular Genetics, 2015, 24, 2861-2872.	1.4	69
23	Biallelic CACNA1A mutations cause early onset epileptic encephalopathy with progressive cerebral, cerebellar, and optic nerve atrophy. American Journal of Medical Genetics, Part A, 2016, 170, 2173-2176.	0.7	65
24	Diffuse hypomyelination is not obligate for POLR3-related disorders. Neurology, 2016, 86, 1622-1626.	1.5	65
25	Three patients with 9p deletions including DMRT1 and DMRT2: A girl with XY complement, bilateral ovotestes, and extreme growth retardation, and two XX females with normal pubertal development. , 2004, 130A, 415-423.		61
26	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
27	De Novo SCN8A Mutation Identified by Whole-Exome Sequencing in a Boy With Neonatal Epileptic Encephalopathy, Multiple Congenital Anomalies, and Movement Disorders. Journal of Child Neurology, 2014, 29, NP202-NP206.	0.7	59
28	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
29	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
30	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	3.6	53
31	Increased Dosage of RAB39B Affects Neuronal Development and Could Explain the Cognitive Impairment in Male Patients with Distal Xq28 Copy Number Gains. Human Mutation, 2014, 35, 377-383.	1.1	52
32	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51
33	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	1.7	50
34	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
35	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	2.6	48
36	DiGeorge/velocardiofacial syndrome: FISH studies of chromosomes 22q11 and 10p14, and clinical reports on the proximal 22q11 deletion. , 2003, 117A, 1-5.		47

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37	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	1.4	47
38	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
39	A novel c.2TÂ>ÂC mutation of the KDM5C/JARID1C gene in one large family with X-linked intellectual disability. European Journal of Medical Genetics, 2012, 55, 178-184.	0.7	42
40	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	1.1	42
41	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	0.5	42
42	Leukoencephalopathy with Calcifications and Cysts: A Purely Neurological Disorder Distinct from Coats Plus. Neuropediatrics, 2014, 45, 175-182.	0.3	41
43	A missense mutation in the catalytic domain of Oâ€GlcNAc transferase links perturbations in protein Oâ€GlcNAcylation to Xâ€linked intellectual disability. FEBS Letters, 2020, 594, 717-727.	1.3	40
44	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
45	An intellectual disability syndrome with single-nucleotide variants in O-GlcNAc transferase. European Journal of Human Genetics, 2020, 28, 706-714.	1.4	38
46	Maternally and paternally inherited deletion of 7q31 involving the FOXP2 gene in two families. American Journal of Medical Genetics, Part A, 2012, 158A, 254-256.	0.7	37
47	Effectiveness of whole exome sequencing in unsolved patients with a clinical suspicion of a mitochondrial disorder in Estonia. Molecular Genetics and Metabolism Reports, 2018, 15, 80-89.	0.4	37
48	Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149.	1.2	36
49	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	2.6	35
50	Prevalence of Angelman syndrome and Praderâ€“Willi syndrome in Estonian children: Sister syndromes not equally represented. American Journal of Medical Genetics, Part A, 2006, 140A, 1936-1943.	0.7	32
51	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. Metabolic Brain Disease, 2018, 33, 875-884.	1.4	32
52	Defective DNA Polymerase Î±-Primase Leads to X-Linked Intellectual Disability Associated with Severe Growth Retardation, Microcephaly, and Hypogonadism. American Journal of Human Genetics, 2019, 104, 957-967.	2.6	32
53	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	1.7	32
54	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	1.1	32

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55	Detection of variants in SLC6A8 and functional analysis of unclassified missense variants. Molecular Genetics and Metabolism, 2012, 105, 596-601.	0.5	31
56	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
57	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome: Opposite Phenotypes with Heterogeneous Molecular Etiology. Molecular Syndromology, 2016, 7, 110-121.	0.3	30
58	Subtelomere FISH in 50 children with mental retardation and minor anomalies, identified by a checklist, detects 10 rearrangements including a de novo balanced translocation of chromosomes 17p13.3 and 20q13.33. American Journal of Medical Genetics Part A, 2004, 128A, 364-373.	2.4	29
59	5.9Mb microdeletion in chromosome band 17q22â€“q23.2 associated with tracheo-esophageal fistula and conductive hearing loss. European Journal of Medical Genetics, 2009, 52, 71-74.	0.7	29
60	The Prevalence of PMM2-CDG in Estonia Based on Population Carrier Frequencies and Diagnosed Patients. JIMD Reports, 2017, 39, 13-17.	0.7	29
61	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. European Journal of Human Genetics, 2018, 26, 407-419.	1.4	29
62	Familial Williams-Beuren syndrome. , 1998, 80, 491-493.		28
63	MECP2mutation analysis in patients with mental retardation. , 2005, 132A, 121-124.		28
64	The neonatal phenotype of Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1241-1244.	0.7	28
65	Novel homozygous mutation in KPTN gene causing a familial intellectual disabilityâ€macrocephaly syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1913-1915.	0.7	28
66	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	9.4	28
67	A Novel Mutation in the SCO2 Gene in a Neonate With Early-Onset Cardioencephalomyopathy. Pediatric Neurology, 2010, 42, 227-230.	1.0	27
68	Investigating the cardiac pathology of SCO2â€mediated hypertrophic cardiomyopathy using patients induced pluripotent stem cellâ€“derived cardiomyocytes. Journal of Cellular and Molecular Medicine, 2018, 22, 913-925.	1.6	27
69	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	1.7	27
70	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	1.4	27
71	Molecular analysis of mucopolysaccharidosis type VI in Poland, Belarus, Lithuania and Estonia. Molecular Genetics and Metabolism, 2012, 105, 237-243.	0.5	26
72	A girl with inverted triplication of chromosome 3q25.3â€‰â†’â€‰q29 and multiple congenital anomalies consistent with 3q duplication syndrome. , 2005, 134A, 434-438.		25

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73	A patient with the classic features of Phelanâ€McDermid syndrome and a high immunoglobulin E level caused by a cryptic interstitial 0.72â€Mb deletion in the 22q13.2 region. American Journal of Medical Genetics, Part A, 2014, 164, 806-809.	0.7	25
74	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	4.7	25
75	Clinical assessment of five patients with BRWD3 mutation at Xq21.1 gives further evidence for mild to moderate intellectual disability and macrocephaly. European Journal of Medical Genetics, 2014, 57, 200-206.	0.7	24
76	Large gene panel sequencing in clinical diagnosticsâ€"results from 501 consecutive cases. Clinical Genetics, 2018, 93, 78-83.	1.0	24
77	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	1.1	24
78	LEOPARD syndrome with recurrent PTPN11 mutation Y279C and different cutaneous manifestations: two case reports and a review of the literature. European Journal of Pediatrics, 2010, 169, 469-473.	1.3	23
79	Long-term complications in Estonian galactosemia patients with a less strict lactose-free diet and metabolic control. Molecular Genetics and Metabolism, 2011, 103, 249-253.	0.5	22
80	Chromosomal microarray analysis as a firstâ€tier clinical diagnostic test: E stonian experience. Molecular Genetics & Genomic Medicine, 2014, 2, 166-175.	0.6	22
81	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. European Journal of Medical Genetics, 2019, 62, 103572.	0.7	22
82	A retrospective analysis of the prevalence of imprinting disorders in Estonia from 1998 to 2016. European Journal of Human Genetics, 2019, 27, 1649-1658.	1.4	21
83	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
84	High incidence of low vitamin B12 levels in Estonian newborns. Molecular Genetics and Metabolism Reports, 2018, 15, 1-5.	0.4	20
85	Prevalence of c.35delG and p.M34T mutations in the GJB2 gene in Estonia. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 1007-1012.	0.4	19
86	The Live-Birth Prevalence of Mucopolysaccharidoses in Estonia. Genetic Testing and Molecular Biomarkers, 2012, 16, 846-849.	0.3	18
87	Coffinâ€"Siris Syndrome with obesity, macrocephaly, hepatomegaly and hyperinsulinism caused by a mutation in the ARID1B gene. European Journal of Human Genetics, 2014, 22, 1327-1329.	1.4	18
88	FLAD1â€associated multiple acylâ€CoA dehydrogenase deficiency identified by newborn screening. Molecular Genetics & Genomic Medicine, 2019, 7, e915.	0.6	18
89	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. Journal of Clinical Investigation, 2021, 131, .	3.9	18
90	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1692-1709.	2.6	18

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91	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
92	Two sisters with Silver-Russell phenotype. , 2004, 131A, 301-306.		16
93	Three families with mild PMM2â€CDG and normal cognitive development. American Journal of Medical Genetics, Part A, 2017, 173, 1620-1624.	0.7	16
94	A new case of 2q duplication supports either a locus for orofacial clefting between markers D2S1897 and D2S2023 or a locus for cleft palate only on chromosome 2q13-q21. American Journal of Medical Genetics, Part A, 2005, 137A, 323-327.	0.7	15
95	Familial 1.3-Mb 11p15.5p15.4 Duplication in Three Generations Causing Silver-Russell and Beckwith-Wiedemann Syndromes. Molecular Syndromology, 2015, 6, 147-151.	0.3	15
96	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	0.7	15
97	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	2.6	15
98	Phenylalanine Hydroxylase Gene Mutation R408W Is Present on 84% of Estonian Phenylketonuria Chromosomes. European Journal of Human Genetics, 1996, 4, 296-300.	1.4	15
99	Recommendations for reporting results of diagnostic genomic testing. European Journal of Human Genetics, 2022, 30, 1011-1016.	1.4	15
100	Congenital disorder of glycosylation caused by starting site-specific variant in syntaxin-5. Nature Communications, 2021, 12, 6227.	5.8	14
101	A New Case of a Rare Combination of Temple Syndrome and Mosaic Trisomy 14 and a Literature Review. Molecular Syndromology, 2018, 9, 182-189.	0.3	13
102	Girl With Partial Turner Syndrome and Absence Epilepsy. Pediatric Neurology, 2008, 38, 289-292.	1.0	12
103	CDKL5 Gene-Related Epileptic Encephalopathy in Estonia: Four Cases, One Novel Mutation Causing Severe Phenotype in a Boy, and Overview of the Literature. Neuropediatrics, 2016, 47, 361-367.	0.3	12
104	Two familial microduplications of 15q26.3 causing overgrowth and variable intellectual disability with normal copy number of IGF1R. European Journal of Medical Genetics, 2016, 59, 257-262.	0.7	12
105	Prevalence of the Fragile X Syndrome Among Estonian Mentally Retarded and the Entire Children's Population. Journal of Child Neurology, 2008, 23, 1400-1405.	0.7	11
106	The Diagnostic Utility of Single Long Contiguous Stretches of Homozygosity in Patients without Parental Consanguinity. Molecular Syndromology, 2015, 6, 135-140.	0.3	11
107	Regulatory landscape of providing information on newborn screening to parents across Europe. European Journal of Human Genetics, 2021, 29, 67-78.	1.4	11
108	Development of the phenylketonuria screening programme in Estonia. Journal of Medical Screening, 1998, 5, 22-23.	1.1	10

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109	Splice variant IVS2-2A>G in the SLC26A5 (Prestin) gene in five Estonian families with hearing loss. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 103-107.	0.4	10
110	Prevalence of Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency in Estonia. JIMD Reports, 2011, 2, 79-85.	0.7	10
111	Patient with Dup(5)(q35.2-q35.3) reciprocal to the common Sotos syndrome deletion and review of the literature. European Journal of Medical Genetics, 2013, 56, 202-206.	0.7	10
112	A prenatally diagnosed case of Meckelâ€“Gruber syndrome with novel compound heterozygous pathogenic variants in the TXNDC15 gene. Molecular Genetics & Genomic Medicine, 2019, 7, e614.	0.6	10
113	Girl with combined cellular immunodeficiency, pancytopenia, malformations, deletion 11q23.3â€‰â†’â€‰qter, and trisomy 8q24.3â€‰â†’â€‰qter. American Journal of Medical Genetics Part A, 2002, 108, 322-326.	2.4	9
114	Hearing impairment in Estonia: An algorithm to investigate genetic causes in pediatric patients. Advances in Medical Sciences, 2013, 58, 419-428.	0.9	9
115	Two Consecutive Pregnancies with Simpson-Golabi-Behmel Syndrome Type 1: Case Report and Review of Published Prenatal Cases. Molecular Syndromology, 2018, 9, 205-213.	0.3	9
116	The Estimated Prevalence of N-Linked Congenital Disorders of Glycosylation Across Various Populations Based on Allele Frequencies in General Population Databases. Frontiers in Genetics, 2021, 12, 719437.	1.1	9
117	A parallel SNP array study of genomic aberrations associated with mental retardation in patients and general population in Estonia. European Journal of Medical Genetics, 2011, 54, 136-143.	0.7	8
118	Monosomy 1p36 â€“ A multifaceted and still enigmatic syndrome: Four clinically diverse cases with shared white matter abnormalities. European Journal of Paediatric Neurology, 2014, 18, 338-346.	0.7	8
119	The Frequency of Methylation Abnormalities Among Estonian Patients Selected by Clinical Diagnostic Scoring Systems for Silverâ€“Russell Syndrome and Beckwithâ€“Wiedemann Syndrome. Genetic Testing and Molecular Biomarkers, 2015, 19, 684-691.	0.3	8
120	De novo exonic mutation in MYH7 gene leading to exon skipping in a patient with early onset muscular weakness and fiber-type disproportion. Neuromuscular Disorders, 2016, 26, 236-239.	0.3	8
121	Genome sequencing identifies a homozygous inversion disrupting QDPR as a cause for dihydropteridine reductase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1154.	0.6	8
122	Descriptive epidemiology of Down's syndrome in Estonia. Paediatric and Perinatal Epidemiology, 2006, 20, 512-519.	0.8	7
123	Characterization of two supernumerary marker chromosomes in a patient with signs of Klinefelter syndrome, mild facial anomalies, and severe speech delay. American Journal of Medical Genetics, Part A, 2006, 140A, 488-495.	0.7	7
124	A Female With Angelman Syndrome and Unusual Limb Deformities. Pediatric Neurology, 2005, 33, 66-69.	1.0	6
125	Classical galactosemia in Estonia: selective neonatal screening, incidence, and genotype/phenotype data of diagnosed patients. Journal of Inherited Metabolic Disease, 2010, 33, 175-176.	1.7	6
126	Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the PRPS1 gene. Molecular Genetics and Metabolism Reports, 2020, 25, 100677.	0.4	6

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127	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	2.6	6
128	Mutation 985A>G in the MCAD gene shows low incidence in Estonian population. Human Mutation, 2000, 15, 293-294.	1.1	5
129	An 8.4â€Mb 3q26.33â€3q28 microdeletion in a patient with blepharophimosisâ€“intellectual disability syndrome and a review of the literature. Clinical Case Reports (discontinued), 2016, 4, 824-830.	0.2	5
130	The incidence and characterization of phenylketonuric patients in Estonia. Journal of Inherited Metabolic Disease, 1996, 19, 381-382.	1.7	4
131	Mosaicism for maternal uniparental disomy 15 in a boy with some clinical features of Praderâ€“Willi syndrome. European Journal of Medical Genetics, 2014, 57, 279-283.	0.7	4
132	Hyperphenylalaninaemias in Estonia: Genotypeâ€“Phenotype Correlation and Comparative Overview of the Patient Cohort Before and After Nation-Wide Neonatal Screening. JIMD Reports, 2017, 40, 39-45.	0.7	4
133	The evaluation of phenylalanine levels in Estonian phenylketonuria patients during eight years by electronic laboratory records. Molecular Genetics and Metabolism Reports, 2019, 19, 100467.	0.4	4
134	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. Human Molecular Genetics, 2020, 29, 1426-1439.	1.4	4
135	CAPN3 c.1746â€20C>G variant is hypomorphic for LGMD R1 calpain 3â€related. Human Mutation, 2022, 43, 1347-1353.	1.1	4
136	Parentsâ€™ Satisfaction with Medical and Social Assistance Provided to Children with Down Syndrome: Experience in Estonia. Public Health Genomics, 2003, 6, 166-170.	0.6	3
137	Prospective experience with contingent screening strategy for Down syndrome in Estonia. Journal of Community Genetics, 2010, 1, 133-138.	0.5	3
138	Incidence of Childhood Epilepsy in Estonia. Journal of Child Neurology, 2018, 33, 587-592.	0.7	3
139	PEHO syndrome caused by compound heterozygote variants in ZNHIT3 gene. European Journal of Medical Genetics, 2020, 63, 103660.	0.7	3
140	The Birth Prevalence of Spinal Muscular Atrophy: A Population Specific Approach in Estonia. Frontiers in Genetics, 2021, 12, 796862.	1.1	3
141	Boy with celiac disease, malformations, and ring chromosome 13 with deletion 13q32?qter. American Journal of Medical Genetics Part A, 2000, 93, 399-402.	2.4	2
142	De novo deletion of HOXB gene cluster in a patient with failure to thrive, developmental delay, gastroesophageal reflux and bronchiectasis. European Journal of Medical Genetics, 2015, 58, 336-340.	0.7	2
143	A twoâ€year prospective study assessing the performance of fetal chromosomal microarray analysis and nextâ€generation sequencing in highâ€risk pregnancies. Molecular Genetics & Genomic Medicine, 2021, 9, e1787.	0.6	2
144	Molecular diagnosis of Down syndrome using quantitative APEXâ€2 microarrays. Prenatal Diagnosis, 2010, 30, 1170-1177.	1.1	1

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145	Periventricular Venous Infarction in an Extremely Premature Infant as the Cause of Schizencephaly. Journal of Pediatric Neurology, 2020, 18, 267-270.	0.0	1
146	Ocular Manifestation of CACNA1A Pathogenic Variants. Pediatric Neurology Briefs, 2016, 30, 46.	0.2	1
147	Complex I deficiency and Leigh syndrome through the eyes of a clinician. EMBO Molecular Medicine, 2020, 12, e13187.	3.3	1