

Katrin Ounap

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

148
papers

4,635
citations

32
h-index

63
g-index

152
ext. papers

5,891
ext. citations

6.1
avg, IF

4.63
L-index

#	Paper	IF	Citations
148	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. <i>Nature</i> , 2010 , 463, 671-5	50.4	403
147	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. <i>Lancet Neurology</i> , 2011 , 10, 806-18	24.1	274
146	Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018 , 14, 229-249	15.2	234
145	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. <i>Journal of Medical Genetics</i> , 2009 , 46, 511-23	5.8	226
144	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017 , 101, 664-685	11	214
143	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. <i>Nature Genetics</i> , 2012 , 44, 338-42	36.3	186
142	A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. <i>Journal of Medical Genetics</i> , 2012 , 49, 660-8	5.8	182
141	Novel (ovario) leukodystrophy related to AARS2 mutations. <i>Neurology</i> , 2014 , 82, 2063-71	6.5	142
140	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. <i>JAMA Psychiatry</i> , 2016 , 73, 20-30	14.5	120
139	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. <i>Journal of Medical Genetics</i> , 2011 , 48, 105-16	5.8	112
138	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. <i>Journal of Medical Genetics</i> , 2009 , 46, 192-7	5.8	112
137	Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. <i>Journal of Medical Genetics</i> , 2013 , 50, 463-72	5.8	89
136	CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. <i>American Journal of Human Genetics</i> , 2015 , 96, 245-57	11	84
135	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 171-176	5.4	83
134	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. <i>Nature Genetics</i> , 2016 , 48, 1185-92	36.3	74
133	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. <i>American Journal of Human Genetics</i> , 2016 , 99, 860-876	11	68
132	Genotype-phenotype correlations in patients with retinoblastoma and interstitial 13q deletions. <i>European Journal of Human Genetics</i> , 2011 , 19, 947-58	5.3	64

131	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016 , 24, 652-9	5.3	57
130	Neurologic phenotypes associated with / mutations: Expanding the spectrum of disease. <i>Neurology</i> , 2018 , 91, e2078-e2088	6.5	55
129	De novo SCN8A mutation identified by whole-exome sequencing in a boy with neonatal epileptic encephalopathy, multiple congenital anomalies, and movement disorders. <i>Journal of Child Neurology</i> , 2014 , 29, NP202-6	2.5	53
128	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, 415-23		53
127	Monogenic variants in dystonia: an exome-wide sequencing study. <i>Lancet Neurology</i> , 2020 , 19, 908-918	11.8	51
126	Biallelic CACNA1A mutations cause early onset epileptic encephalopathy with progressive cerebral, cerebellar, and optic nerve atrophy. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2173-6	2.5	50
125	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 5-28	5.4	45
124	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. <i>Human Molecular Genetics</i> , 2014 , 23, 6069-80	5.6	45
123	Diffuse hypomyelination is not obligate for POLR3-related disorders. <i>Neurology</i> , 2016 , 86, 1622-6	6.5	42
122	Increased dosage of RAB39B affects neuronal development and could explain the cognitive impairment in male patients with distal Xq28 copy number gains. <i>Human Mutation</i> , 2014 , 35, 377-83	4.7	40
121	DiGeorge/velocardiofacial syndrome: FISH studies of chromosomes 22q11 and 10p14, and clinical reports on the proximal 22q11 deletion 2003 , 117A, 1-5		38
120	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , 2015 , 96, 784-96	11	35
119	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. <i>Human Molecular Genetics</i> , 2015 , 24, 2861-72	5.6	33
118	Leukoencephalopathy with calcifications and cysts: a purely neurological disorder distinct from coats plus. <i>Neuropediatrics</i> , 2014 , 45, 175-82	1.6	33
117	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. <i>Fertility and Sterility</i> , 2017 , 108, 168-174	4.8	32
116	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. <i>Journal of Inherited Metabolic Disease</i> , 2011 , 34, 923-7	5.4	31
115	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. <i>American Journal of Human Genetics</i> , 2019 , 105, 493-508	11	30
114	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018 , 102, 744-759	11	30

113	A novel c.2T>C mutation of the KDM5C/JARID1C gene in one large family with X-linked intellectual disability. <i>European Journal of Medical Genetics</i> , 2012 , 55, 178-84	2.6	27
112	MECP2 mutation analysis in patients with mental retardation 2005 , 132A, 121-4		27
111	Effectiveness of whole exome sequencing in unsolved patients with a clinical suspicion of a mitochondrial disorder in Estonia. <i>Molecular Genetics and Metabolism Reports</i> , 2018 , 15, 80-89	1.8	26
110	5.9 Mb microdeletion in chromosome band 17q22-q23.2 associated with tracheo-esophageal fistula and conductive hearing loss. <i>European Journal of Medical Genetics</i> , 2009 , 52, 71-4	2.6	26
109	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. <i>European Journal of Human Genetics</i> , 2019 , 27, 747-759	5.3	25
108	Maternally and paternally inherited deletion of 7q31 involving the FOXP2 gene in two families. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 254-6	2.5	25
107	Molecular analysis of mucopolysaccharidosis type VI in Poland, Belarus, Lithuania and Estonia. <i>Molecular Genetics and Metabolism</i> , 2012 , 105, 237-43	3.7	25
106	A novel mutation in the SCO2 gene in a neonate with early-onset cardioencephalomyopathy. <i>Pediatric Neurology</i> , 2010 , 42, 227-30	2.9	25
105	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. <i>American Journal of Medical Genetics Part A</i> , 2004 , 128A, 364-73		25
104	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. <i>Genetics in Medicine</i> , 2020 , 22, 1102-1107	8.1	24
103	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome: Opposite Phenotypes with Heterogeneous Molecular Etiology. <i>Molecular Syndromology</i> , 2016 , 7, 110-21	1.5	24
102	Detection of variants in SLC6A8 and functional analysis of unclassified missense variants. <i>Molecular Genetics and Metabolism</i> , 2012 , 105, 596-601	3.7	23
101	A girl with inverted triplication of chromosome 3q25.3 --> q29 and multiple congenital anomalies consistent with 3q duplication syndrome 2005 , 134, 434-8		23
100	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. <i>Clinical Epigenetics</i> , 2020 , 12, 7	7.7	23
99	The neonatal phenotype of Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 1241-4	2.5	22
98	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. <i>American Journal of Human Genetics</i> , 2020 , 106, 234-245	11	22
97	A patient with the classic features of Phelan-McDermid syndrome and a high immunoglobulin E level caused by a cryptic interstitial 0.72-Mb deletion in the 22q13.2 region. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 806-9	2.5	21
96	The Prevalence of PMM2-CDG in Estonia Based on Population Carrier Frequencies and Diagnosed Patients. <i>JIMD Reports</i> , 2018 , 39, 13-17	1.9	21

95	LEOPARD syndrome with recurrent PTPN11 mutation Y279C and different cutaneous manifestations: two case reports and a review of the literature. <i>European Journal of Pediatrics</i> , 2010 , 169, 469-73	4.1	21
94	Prevalence of Angelman syndrome and Prader-Willi syndrome in Estonian children: sister syndromes not equally represented. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 1936-43	2.5	21
93	Clinical, neuroradiological, and biochemical features of SLC35A2-CDG patients. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 553-564	5.4	21
92	Can untreated PKU patients escape from intellectual disability? A systematic review. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 149	4.2	21
91	Large gene panel sequencing in clinical diagnostics-results from 501 consecutive cases. <i>Clinical Genetics</i> , 2018 , 93, 78-83	4	20
90	Long-term complications in Estonian galactosemia patients with a less strict lactose-free diet and metabolic control. <i>Molecular Genetics and Metabolism</i> , 2011 , 103, 249-53	3.7	19
89	Prevalence of c.35delG and p.M34T mutations in the GJB2 gene in Estonia. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2010 , 74, 1007-12	1.7	19
88	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018 , 50, 1442-1451	36.3	19
87	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. <i>Genetics in Medicine</i> , 2019 , 21, 2723-2733	8.1	18
86	Novel homozygous mutation in KPTN gene causing a familial intellectual disability-macrocephaly syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1913-5	2.5	18
85	Familial Williams-Beuren syndrome. <i>American Journal of Medical Genetics Part A</i> , 1998 , 80, 491-3		18
84	Defective DNA Polymerase ϵ Primase Leads to X-Linked Intellectual Disability Associated with Severe Growth Retardation, Microcephaly, and Hypogonadism. <i>American Journal of Human Genetics</i> , 2019 , 104, 957-967	11	17
83	An intellectual disability syndrome with single-nucleotide variants in O-GlcNAc transferase. <i>European Journal of Human Genetics</i> , 2020 , 28, 706-714	5.3	17
82	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019 , 105, 403-412	11	17
81	Chromosomal microarray analysis as a first-tier clinical diagnostic test: Estonian experience. <i>Molecular Genetics & Genomic Medicine</i> , 2014 , 2, 166-75	2.3	17
80	A missense mutation in the catalytic domain of O-GlcNAc transferase links perturbations in protein O-GlcNAcylation to X-linked intellectual disability. <i>FEBS Letters</i> , 2020 , 594, 717-727	3.8	17
79	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. <i>Metabolic Brain Disease</i> , 2018 , 33, 875-884	3.9	16
78	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. <i>European Journal of Human Genetics</i> , 2018 , 26, 407-419	5.3	15

77	Investigating the cardiac pathology of SCO2-mediated hypertrophic cardiomyopathy using patients induced pluripotent stem cell-derived cardiomyocytes. <i>Journal of Cellular and Molecular Medicine</i> , 2018 , 22, 913-925	5.6	15
76	Coffin-Siris Syndrome with obesity, macrocephaly, hepatomegaly and hyperinsulinism caused by a mutation in the ARID1B gene. <i>European Journal of Human Genetics</i> , 2014 , 22, 1327-9	5.3	15
75	The live-birth prevalence of mucopolysaccharidoses in Estonia. <i>Genetic Testing and Molecular Biomarkers</i> , 2012 , 16, 846-9	1.6	15
74	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. <i>European Journal of Human Genetics</i> , 2020 , 28, 770-782	5.3	13
73	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 853-860	5.4	13
72	Clinical assessment of five patients with BRWD3 mutation at Xq21.1 gives further evidence for mild to moderate intellectual disability and macrocephaly. <i>European Journal of Medical Genetics</i> , 2014 , 57, 200-6	2.6	12
71	Familial 1.3-Mb 11p15.5p15.4 Duplication in Three Generations Causing Silver-Russell and Beckwith-Wiedemann Syndromes. <i>Molecular Syndromology</i> , 2015 , 6, 147-51	1.5	12
70	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020 , 6,	14.3	12
69	Three families with mild PMM2-CDG and normal cognitive development. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1620-1624	2.5	11
68	High incidence of low vitamin B12 levels in Estonian newborns. <i>Molecular Genetics and Metabolism Reports</i> , 2018 , 15, 1-5	1.8	11
67	A New Case of a Rare Combination of Temple Syndrome and Mosaic Trisomy 14 and a Literature Review. <i>Molecular Syndromology</i> , 2018 , 9, 182-189	1.5	11
66	FLAD1-associated multiple acyl-CoA dehydrogenase deficiency identified by newborn screening. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e915	2.3	11
65	Two sisters with Silver-Russell phenotype 2004 , 131, 301-6		11
64	Phenylalanine hydroxylase gene mutation R408W is present on 84% of Estonian phenylketonuria chromosomes. <i>European Journal of Human Genetics</i> , 1996 , 4, 296-300	5.3	11
63	A retrospective analysis of the prevalence of imprinting disorders in Estonia from 1998 to 2016. <i>European Journal of Human Genetics</i> , 2019 , 27, 1649-1658	5.3	10
62	Prevalence of the fragile X syndrome among Estonian mentally retarded and the entire children population. <i>Journal of Child Neurology</i> , 2008 , 23, 1400-5	2.5	10
61	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. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 137A, 323-7	2.5	10
60	CDKL5 Gene-Related Epileptic Encephalopathy in Estonia: Four Cases, One Novel Mutation Causing Severe Phenotype in a Boy, and Overview of the Literature. <i>Neuropediatrics</i> , 2016 , 47, 361-367	1.6	10

59	Girl with partial Turner syndrome and absence epilepsy. <i>Pediatric Neurology</i> , 2008 , 38, 289-92	2.9	9
58	Development of the phenylketonuria screening programme in Estonia. <i>Journal of Medical Screening</i> , 1998 , 5, 22-3	1.4	9
57	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. <i>Genetics in Medicine</i> , 2021 , 23, 543-554	8.1	9
56	Two familial microduplications of 15q26.3 causing overgrowth and variable intellectual disability with normal copy number of IGF1R. <i>European Journal of Medical Genetics</i> , 2016 , 59, 257-62	2.6	8
55	Patient with dup(5)(q35.2-q35.3) reciprocal to the common Sotos syndrome deletion and review of the literature. <i>European Journal of Medical Genetics</i> , 2013 , 56, 202-6	2.6	8
54	The Frequency of Methylation Abnormalities Among Estonian Patients Selected by Clinical Diagnostic Scoring Systems for Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome. <i>Genetic Testing and Molecular Biomarkers</i> , 2015 , 19, 684-91	1.6	8
53	Splice variant IVS2-2A>G in the SLC26A5 (Prestin) gene in five Estonian families with hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2009 , 73, 103-7	1.7	8
52	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. <i>European Journal of Medical Genetics</i> , 2019 , 62, 103572	2.6	8
51	A prenatally diagnosed case of Meckel-Gruber syndrome with novel compound heterozygous pathogenic variants in the TXNDC15 gene. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e614	2.3	7
50	Monosomy 1p36 - a multifaceted and still enigmatic syndrome: four clinically diverse cases with shared white matter abnormalities. <i>European Journal of Paediatric Neurology</i> , 2014 , 18, 338-46	3.8	7
49	Hearing impairment in Estonia: an algorithm to investigate genetic causes in pediatric patients. <i>Advances in Medical Sciences</i> , 2013 , 58, 419-28	2.8	7
48	The Diagnostic Utility of Single Long Contiguous Stretches of Homozygosity in Patients without Parental Consanguinity. <i>Molecular Syndromology</i> , 2015 , 6, 135-40	1.5	7
47	A parallel SNP array study of genomic aberrations associated with mental retardation in patients and general population in Estonia. <i>European Journal of Medical Genetics</i> , 2011 , 54, 136-43	2.6	7
46	Descriptive epidemiology of Down's syndrome in Estonia. <i>Paediatric and Perinatal Epidemiology</i> , 2006 , 20, 512-9	2.7	7
45	Girl with combined cellular immunodeficiency, pancytopenia, malformations, deletion 11q23.3 --> qter, and trisomy 8q24.3 --> qter. <i>American Journal of Medical Genetics Part A</i> , 2002 , 108, 322-6		7
44	Prevalence of Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency in Estonia. <i>JIMD Reports</i> , 2012 , 2, 79-85	1.9	6
43	A female with Angelman syndrome and unusual limb deformities. <i>Pediatric Neurology</i> , 2005 , 33, 66-9	2.9	6
42	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 119-133	2.5	6

41	De novo exonic mutation in MYH7 gene leading to exon skipping in a patient with early onset muscular weakness and fiber-type disproportion. <i>Neuromuscular Disorders</i> , 2016 , 26, 236-9	2.9	5
40	Classical galactosemia in Estonia: selective neonatal screening, incidence, and genotype/phenotype data of diagnosed patients. <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33, 175-6	5.4	5
39	Characterization of two supernumerary marker chromosomes in a patient with signs of Klinefelter syndrome, mild facial anomalies, and severe speech delay. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 488-95	2.5	5
38	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 15-25	2.5	5
37	Hyperphenylalaninaemias in Estonia: Genotype-Phenotype Correlation and Comparative Overview of the Patient Cohort Before and After Nation-Wide Neonatal Screening. <i>JIMD Reports</i> , 2018 , 40, 39-45	1.9	4
36	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. <i>Genome Medicine</i> , 2021 , 13, 153	14.4	4
35	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. <i>Genetics in Medicine</i> , 2021 , 23, 1474-1483	8.1	4
34	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. <i>Science Advances</i> , 2021 , 7,	14.3	4
33	Genome sequencing identifies a homozygous inversion disrupting QDPR as a cause for dihydropteridine reductase deficiency. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1154	2.3	3
32	Mosaicism for maternal uniparental disomy 15 in a boy with some clinical features of Prader-Willi syndrome. <i>European Journal of Medical Genetics</i> , 2014 , 57, 279-83	2.6	3
31	Parents' satisfaction with medical and social assistance provided to children with Down syndrome: experience in Estonia. <i>Public Health Genomics</i> , 2003 , 6, 166-70	1.9	3
30	The incidence and characterization of phenylketonuric patients in Estonia. <i>Journal of Inherited Metabolic Disease</i> , 1996 , 19, 381-2	5.4	3
29	A form of muscular dystrophy associated with pathogenic variants in JAG2. <i>American Journal of Human Genetics</i> , 2021 , 108, 840-856	11	3
28	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021 , 108, 1053-1068	11	3
27	An 8.4-Mb 3q26.33-3q28 microdeletion in a patient with blepharophimosis-intellectual disability syndrome and a review of the literature. <i>Clinical Case Reports (discontinued)</i> , 2016 , 4, 824-30	0.7	3
26	Regulatory landscape of providing information on newborn screening to parents across Europe. <i>European Journal of Human Genetics</i> , 2021 , 29, 67-78	5.3	3
25	POLRMT mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021 , 12, 1135	17.4	3
24	Two Consecutive Pregnancies with Simpson-Golabi-Behmel Syndrome Type 1: Case Report and Review of Published Prenatal Cases. <i>Molecular Syndromology</i> , 2018 , 9, 205-213	1.5	3

23	The evaluation of phenylalanine levels in Estonian phenylketonuria patients during eight years by electronic laboratory records. <i>Molecular Genetics and Metabolism Reports</i> , 2019 , 19, 100467	1.8	2
22	Boy with celiac disease, malformations, and ring chromosome 13 with deletion 13q32->qter. <i>American Journal of Medical Genetics Part A</i> , 2000 , 93, 399-402		2
21	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. <i>Journal of Clinical Investigation</i> , 2021 , 131,	15.9	2
20	Incidence of Childhood Epilepsy in Estonia. <i>Journal of Child Neurology</i> , 2018 , 33, 587-592	2.5	2
19	The Estimated Prevalence of N-Linked Congenital Disorders of Glycosylation Across Various Populations Based on Allele Frequencies in General Population Databases. <i>Frontiers in Genetics</i> , 2021 , 12, 719437	4.5	2
18	A two-year prospective study assessing the performance of fetal chromosomal microarray analysis and next-generation sequencing in high-risk pregnancies. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1787	2.3	2
17	Prospective experience with contingent screening strategy for Down syndrome in Estonia. <i>Journal of Community Genetics</i> , 2010 , 1, 133-8	2.5	1
16	Molecular diagnosis of Down syndrome using quantitative APEX-2 microarrays. <i>Prenatal Diagnosis</i> , 2010 , 30, 1170-7	3.2	1
15	Mutation 985A>G in the MCAD gene shows low incidence in Estonian population. <i>Human Mutation</i> , 2000 , 15, 293-4	4.7	1
14	Congenital disorder of glycosylation caused by starting site-specific variant in syntaxin-5		1
13	Bi-allelic variants in TSPOAP1, encoding the active zone protein RIMBP1, cause autosomal recessive dystonia		1
12	Identifying long indels in exome sequencing data of patients with intellectual disability		1
11	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2021 , 108, 749-756	11	1
10	PEHO syndrome caused by compound heterozygote variants in ZNHIT3 gene. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103660	2.6	1
9	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021 , 108, 1692-1709	11	1
8	Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the gene. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 25, 100677	1.8	0
7	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. <i>Human Molecular Genetics</i> , 2020 , 29, 1426-1439	5.6	0
6	Congenital disorder of glycosylation caused by starting site-specific variant in syntaxin-5. <i>Nature Communications</i> , 2021 , 12, 6227	17.4	0

- 5 The Birth Prevalence of Spinal Muscular Atrophy: A Population Specific Approach in Estonia.. *Frontiers in Genetics*, **2021**, 12, 796862 4.5 ○
- 4 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, 336-40 2.6
- 3 Complex I deficiency and Leigh syndrome through the eyes of a clinician. *EMBO Molecular Medicine*, **2020**, 12, e13187 12
- 2 Ocular Manifestation of Pathogenic Variants. *Pediatric Neurology Briefs*, **2016**, 30, 46 0.3
- 1 Periventricular Venous Infarction in an Extremely Premature Infant as the Cause of Schizencephaly. *Journal of Pediatric Neurology*, **2020**, 18, 267-270 0.2