Katrin Ounap

List of Publications by Year in Descending Order

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	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. *Genome Medicine*, **2021**, 13, 153	14.4	4
147	Congenital disorder of glycosylation caused by starting site-specific variant in syntaxin-5. *Nature Communications*, **2021**, 12, 6227	17.4	0
146	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. *Journal of Clinical Investigation*, **2021**, 131,	15.9	2
145	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	11	1
144	A form of muscular dystrophy associated with pathogenic variants in JAG2. *American Journal of Human Genetics*, **2021**, 108, 840-856	11	3
143	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	8.1	4
142	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	11	3
141	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	8.1	9
140	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2021**, 185, 15-25	2.5	5
139	Regulatory landscape of providing information on newborn screening to parents across Europe. *European Journal of Human Genetics*, **2021**, 29, 67-78	5.3	3
138	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 119-133	2.5	6
137	POLRMT mutations impair mitochondrial transcription causing neurological disease. *Nature Communications*, **2021**, 12, 1135	17.4	3
136	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	4.5	2
135	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. *American Journal of Human Genetics*, **2021**, 108, 1692-1709	11	1
134	A two-year prospective study assessing the performance of fetal chromosomal microarray analysis and next-generation sequencing in high-risk pregnancies. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1787	2.3	2
133	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. *Science Advances*, **2021**, 7,	14.3	4
132	The Birth Prevalence of Spinal Muscular Atrophy: A Population Specific Approach in Estonia.. *Frontiers in Genetics*, **2021**, 12, 796862	4.5	0

131	Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the gene. *Molecular Genetics and Metabolism Reports*, **2020**, 25, 100677	1.8	0
130	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	5.6	0
129	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. *Genetics in Medicine*, **2020**, 22, 1102-1107	8.1	24
128	An intellectual disability syndrome with single-nucleotide variants in O-GlcNAc transferase. *European Journal of Human Genetics*, **2020**, 28, 706-714	5.3	17
127	Genome sequencing identifies a homozygous inversion disrupting QDPR as a cause for dihydropteridine reductase deficiency. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1154	2.3	3
126	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	5.3	13
125	Complex I deficiency and Leigh syndrome through the eyes of a clinician. *EMBO Molecular Medicine*, **2020**, 12, e13187	12	
124	A missense mutation in the catalytic domain of O-GlcNAc transferase links perturbations in protein O-GlcNAcylation to X-linked intellectual disability. *FEBS Letters*, **2020**, 594, 717-727	3.8	17
123	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. *American Journal of Human Genetics*, **2020**, 106, 234-245	11	22
122	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
121	Monogenic variants in dystonia: an exome-wide sequencing study. *Lancet Neurology, The*, **2020**, 19, 908-918	24.1	51
120	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
119	Periventricular Venous Infarction in an Extremely Premature Infant as the Cause of Schizencephaly. *Journal of Pediatric Neurology*, **2020**, 18, 267-270	0.2	
118	PEHO syndrome caused by compound heterozygote variants in ZNHIT3 gene. *European Journal of Medical Genetics*, **2020**, 63, 103660	2.6	1
117	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	11	30
116	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	5.3	25
115	Defective DNA Polymerase Primase Leads to X-Linked Intellectual Disability Associated with Severe Growth Retardation, Microcephaly, and Hypogonadism. *American Journal of Human Genetics* , **2019**, 104, 957-967	11	17
114	A prenatally diagnosed case of Meckel-Gruber syndrome with novel compound heterozygous pathogenic variants in the TXNDC15 gene. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e614	2.3	7

113	The evaluation of phenylalanine levels in Estonian phenylketonuria patients during eight years by electronic laboratory records. *Molecular Genetics and Metabolism Reports*, **2019**, 19, 100467	1.8	2
112	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. *Journal of Inherited Metabolic Disease*, **2019**, 42, 5-28	5.4	45
111	FLAD1-associated multiple acyl-CoA dehydrogenase deficiency identified by newborn screening. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e915	2.3	11
110	A retrospective analysis of the prevalence of imprinting disorders in Estonia from 1998 to 2016. *European Journal of Human Genetics*, **2019**, 27, 1649-1658	5.3	10
109	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 105, 403-412	11	17
108	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. *Genetics in Medicine*, **2019**, 21, 2723-2733	8.1	18
107	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. *European Journal of Medical Genetics*, **2019**, 62, 103572	2.6	8
106	Clinical, neuroradiological, and biochemical features of SLC35A2-CDG patients. *Journal of Inherited Metabolic Disease*, **2019**, 42, 553-564	5.4	21
105	Hyperphenylalaninaemias in Estonia: Genotype-Phenotype Correlation and Comparative Overview of the Patient Cohort Before and After Nation-Wide Neonatal Screening. *JIMD Reports*, **2018**, 40, 39-45	1.9	4
104	Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. *Nature Reviews Endocrinology*, **2018**, 14, 229-249	15.2	234
103	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. *European Journal of Human Genetics*, **2018**, 26, 407-419	5.3	15
102	High incidence of low vitamin B12 levels in Estonian newborns. *Molecular Genetics and Metabolism Reports*, **2018**, 15, 1-5	1.8	11
101	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. *Metabolic Brain Disease*, **2018**, 33, 875-884	3.9	16
100	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	11	30
99	Large gene panel sequencing in clinical diagnostics-results from 501 consecutive cases. *Clinical Genetics*, **2018**, 93, 78-83	4	20
98	A New Case of a Rare Combination of Temple Syndrome and Mosaic Trisomy 14 and a Literature Review. *Molecular Syndromology*, **2018**, 9, 182-189	1.5	11
97	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	1.8	26
96	Investigating the cardiac pathology of SCO2-mediated hypertrophic cardiomyopathy using patients induced pluripotent stem cell-derived cardiomyocytes. *Journal of Cellular and Molecular Medicine*, **2018**, 22, 913-925	5.6	15

95	The Prevalence of PMM2-CDG in Estonia Based on Population Carrier Frequencies and Diagnosed Patients. *JIMD Reports*, **2018**, 39, 13-17	1.9	21
94	Neurologic phenotypes associated with / mutations: Expanding the spectrum of disease. *Neurology*, **2018**, 91, e2078-e2088	6.5	55
93	Can untreated PKU patients escape from intellectual disability? A systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 149	4.2	21
92	Two Consecutive Pregnancies with Simpson-Golabi-Behmel Syndrome Type 1: Case Report and Review of Published Prenatal Cases. *Molecular Syndromology*, **2018**, 9, 205-213	1.5	3
91	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. *Nature Genetics*, **2018**, 50, 1442-1451	36.3	19
90	Incidence of Childhood Epilepsy in Estonia. *Journal of Child Neurology*, **2018**, 33, 587-592	2.5	2
89	Three families with mild PMM2-CDG and normal cognitive development. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1620-1624	2.5	11
88	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
87	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	5.4	13
86	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. *Journal of Inherited Metabolic Disease*, **2017**, 40, 171-176	5.4	83
85	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. *Fertility and Sterility*, **2017**, 108, 168-174	4.8	32
84	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, 652-9	5.3	57
83	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92	36.3	74
82	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome: Opposite Phenotypes with Heterogeneous Molecular Etiology. *Molecular Syndromology*, **2016**, 7, 110-21	1.5	24
81	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, 236-9	2.9	5
80	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, 257-62	2.6	8
79	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
78	Ocular Manifestation of Pathogenic Variants. *Pediatric Neurology Briefs*, **2016**, 30, 46	0.3	

77	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, 2173-6	2.5	50
76	Diffuse hypomyelination is not obligate for POLR3-related disorders. *Neurology*, **2016**, 86, 1622-6	6.5	42
75	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	1.6	10
74	An 8.4-Mb 3q26.33-3q28 microdeletion in a patient with blepharophimosis-intellectual disability syndrome and a review of the literature. *Clinical Case Reports (discontinued)*, **2016**, 4, 824-30	0.7	3
73	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	11	68
72	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. *Human Molecular Genetics*, **2015**, 24, 2861-72	5.6	33
71	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	
70	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
69	Novel homozygous mutation in KPTN gene causing a familial intellectual disability-macrocephaly syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1913-5	2.5	18
68	Familial 1.3-Mb 11p15.5p15.4 Duplication in Three Generations Causing Silver-Russell and Beckwith-Wiedemann Syndromes. *Molecular Syndromology*, **2015**, 6, 147-51	1.5	12
67	The Diagnostic Utility of Single Long Contiguous Stretches of Homozygosity in Patients without Parental Consanguinity. *Molecular Syndromology*, **2015**, 6, 135-40	1.5	7
66	The Frequency of Methylation Abnormalities Among Estonian Patients Selected by Clinical Diagnostic Scoring Systems for Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome. *Genetic Testing and Molecular Biomarkers*, **2015**, 19, 684-91	1.6	8
65	CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. *American Journal of Human Genetics*, **2015**, 96, 245-57	11	84
64	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. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 806-9	2.5	21
63	Mosaicism for maternal uniparental disomy 15 in a boy with some clinical features of Prader-Willi syndrome. *European Journal of Medical Genetics*, **2014**, 57, 279-83	2.6	3
62	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, 200-6	2.6	12
61	Monosomy 1p36 - a multifaceted and still enigmatic syndrome: four clinically diverse cases with shared white matter abnormalities. *European Journal of Paediatric Neurology*, **2014**, 18, 338-46	3.8	7
60	Novel (ovario) leukodystrophy related to AARS2 mutations. *Neurology*, **2014**, 82, 2063-71	6.5	142

59	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
58	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, NP202-6	2.5	53
57	Coffin-Siris Syndrome with obesity, macrocephaly, hepatomegaly and hyperinsulinism caused by a mutation in the ARID1B gene. *European Journal of Human Genetics*, **2014**, 22, 1327-9	5.3	15
56	Leukoencephalopathy with calcifications and cysts: a purely neurological disorder distinct from coats plus. *Neuropediatrics*, **2014**, 45, 175-82	1.6	33
55	Chromosomal microarray analysis as a first-tier clinical diagnostic test: Estonian experience. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 166-75	2.3	17
54	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, 377-83	4.7	40
53	Hearing impairment in Estonia: an algorithm to investigate genetic causes in pediatric patients. *Advances in Medical Sciences*, **2013**, 58, 419-28	2.8	7
52	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, 202-6	2.6	8
51	Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. *Journal of Medical Genetics*, **2013**, 50, 463-72	5.8	89
50	Maternally and paternally inherited deletion of 7q31 involving the FOXP2 gene in two families. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 254-6	2.5	25
49	The live-birth prevalence of mucopolysaccharidoses in Estonia. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 846-9	1.6	15
48	Molecular analysis of mucopolysaccharidosis type VI in Poland, Belarus, Lithuania and Estonia. *Molecular Genetics and Metabolism*, **2012**, 105, 237-43	3.7	25
47	Detection of variants in SLC6A8 and functional analysis of unclassified missense variants. *Molecular Genetics and Metabolism*, **2012**, 105, 596-601	3.7	23
46	A novel c.2T□C mutation of the KDM5C/JARID1C gene in one large family with X-linked intellectual disability. *European Journal of Medical Genetics*, **2012**, 55, 178-84	2.6	27
45	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42	36.3	186
44	Prevalence of Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency in Estonia. *JIMD Reports*, **2012**, 2, 79-85	1.9	6
43	A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *Journal of Medical Genetics*, **2012**, 49, 660-8	5.8	182
42	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. *Journal of Medical Genetics*, **2011**, 48, 105-16	5.8	112

41	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, 136-43	2.6	7
40	Long-term complications in Estonian galactosemia patients with a less strict lactose-free diet and metabolic control. *Molecular Genetics and Metabolism*, **2011**, 103, 249-53	3.7	19
39	Genotype-phenotype correlations in patients with retinoblastoma and interstitial 13q deletions. *European Journal of Human Genetics*, **2011**, 19, 947-58	5.3	64
38	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. *Lancet Neurology, The*, **2011**, 10, 806-18	24.1	274
37	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. *Journal of Inherited Metabolic Disease*, **2011**, 34, 923-7	5.4	31
36	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. *Nature*, **2010**, 463, 671-5	50.4	403
35	Prevalence of c.35delG and p.M34T mutations in the GJB2 gene in Estonia. *International Journal of Pediatric Otorhinolaryngology*, **2010**, 74, 1007-12	1.7	19
34	A novel mutation in the SCO2 gene in a neonate with early-onset cardioencephalomyopathy. *Pediatric Neurology*, **2010**, 42, 227-30	2.9	25
33	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, 469-73	4.1	21
32	Prospective experience with contingent screening strategy for Down syndrome in Estonia. *Journal of Community Genetics*, **2010**, 1, 133-8	2.5	1
31	Classical galactosemia in Estonia: selective neonatal screening, incidence, and genotype/phenotype data of diagnosed patients. *Journal of Inherited Metabolic Disease*, **2010**, 33, 175-6	5.4	5
30	Molecular diagnosis of Down syndrome using quantitative APEX-2 microarrays. *Prenatal Diagnosis*, **2010**, 30, 1170-7	3.2	1
29	5.9 Mb microdeletion in chromosome band 17q22-q23.2 associated with tracheo-esophageal fistula and conductive hearing loss. *European Journal of Medical Genetics*, **2009**, 52, 71-4	2.6	26
28	Splice variant IVS2-2A>G in the SLC26A5 (Prestin) gene in five Estonian families with hearing loss. *International Journal of Pediatric Otorhinolaryngology*, **2009**, 73, 103-7	1.7	8
27	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, 511-23	5.8	226
26	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*, **2009**, 46, 192-7	5.8	112
25	Girl with partial Turner syndrome and absence epilepsy. *Pediatric Neurology*, **2008**, 38, 289-92	2.9	9
24	Prevalence of the fragile X syndrome among Estonian mentally retarded and the entire children population. *Journal of Child Neurology*, **2008**, 23, 1400-5	2.5	10

23	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**, 140, 488-95	2.5	5
22	The neonatal phenotype of Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1241-4	2.5	22
21	Prevalence of Angelman syndrome and Prader-Willi syndrome in Estonian children: sister syndromes not equally represented. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1936-43	2.5	21
20	Descriptive epidemiology of Down syndrome in Estonia. *Paediatric and Perinatal Epidemiology*, **2006**, 20, 512-9	2.7	7
19	A female with Angelman syndrome and unusual limb deformities. *Pediatric Neurology*, **2005**, 33, 66-9	2.9	6
18	A girl with inverted triplication of chromosome 3q25.3 --> q29 and multiple congenital anomalies consistent with 3q duplication syndrome **2005**, 134, 434-8		23
17	MECP2 mutation analysis in patients with mental retardation **2005**, 132A, 121-4		27
16	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, 323-7	2.5	10
15	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, 364-73		25
14	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
13	Two sisters with Silver-Russell phenotype **2004**, 131, 301-6		11
12	DiGeorge/velocardiofacial syndrome: FISH studies of chromosomes 22q11 and 10p14, and clinical reports on the proximal 22q11 deletion **2003**, 117A, 1-5		38
11	Parents satisfaction with medical and social assistance provided to children with Down syndrome: experience in Estonia. *Public Health Genomics*, **2003**, 6, 166-70	1.9	3
10	Girl with combined cellular immunodeficiency, pancytopenia, malformations, deletion 11q23.3 --> qter, and trisomy 8q24.3 --> qter. *American Journal of Medical Genetics Part A*, **2002**, 108, 322-6		7
9	Mutation 985A>G in the MCAD gene shows low incidence in Estonian population. *Human Mutation*, **2000**, 15, 293-4	4.7	1
8	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
7	Familial Williams-Beuren syndrome. *American Journal of Medical Genetics Part A*, **1998**, 80, 491-3		18
6	Development of the phenylketonuria screening programme in Estonia. *Journal of Medical Screening* , **1998**, 5, 22-3	1.4	9

5	The incidence and characterization of phenylketonuric patients in Estonia. *Journal of Inherited Metabolic Disease*, **1996**, 19, 381-2	5.4	3
4	Phenylalanine hydroxylase gene mutation R408W is present on 84% of Estonian phenylketonuria chromosomes. *European Journal of Human Genetics*, **1996**, 4, 296-300	5.3	11
3	Congenital disorder of glycosylation caused by starting site-specific variant in syntaxin-5		1
2	Bi-allelic variants inTSPOAP1, encoding the active zone protein RIMBP1, cause autosomal recessive dystonia		1
1	Identifying long indels in exome sequencing data of patients with intellectual disability		1