

# Hanka Venselaar

## List of Publications by Year in descending order

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116  
papers

7,210  
citations

70961

41  
h-index

64668

79  
g-index

121  
all docs

121  
docs citations

121  
times ranked

13470  
citing authors

#	ARTICLE	IF	CITATIONS
1	Protein structure analysis of mutations causing inheritable diseases. An e-Science approach with life scientist friendly interfaces. <i>BMC Bioinformatics</i> , 2010, 11, 548.	1.2	819
2	Human Dectin-1 Deficiency and Mucocutaneous Fungal Infections. <i>New England Journal of Medicine</i> , 2009, 361, 1760-1767.	13.9	671
3	Mutations in SMAD3 cause a syndromic form of aortic aneurysms and dissections with early-onset osteoarthritis. <i>Nature Genetics</i> , 2011, 43, 121-126.	9.4	583
4	Mutations in a TGF- $\beta$ Ligand, TGFB3, Cause Syndromic Aortic Aneurysms and Dissections. <i>Journal of the American College of Cardiology</i> , 2015, 65, 1324-1336.	1.2	238
5	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
6	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. <i>Journal of Medical Genetics</i> , 2009, 46, 598-606.	1.5	194
7	Dominant missense mutations in ABCC9 cause Cantu's syndrome. <i>Nature Genetics</i> , 2012, 44, 793-796.	9.4	184
8	Acyl-CoA Dehydrogenase 9 Is Required for the Biogenesis of Oxidative Phosphorylation Complex I. <i>Cell Metabolism</i> , 2010, 12, 283-294.	7.2	172
9	Mutations in NDUFAF3 (C3ORF60), Encoding an NDUFAF4 (C6ORF66)-Interacting Complex I Assembly Protein, Cause Fatal Neonatal Mitochondrial Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 718-727.	2.6	155
10	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. <i>American Journal of Human Genetics</i> , 2008, 82, 125-138.	2.6	127
11	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. <i>Human Mutation</i> , 2010, 31, 656-666.	1.1	126
12	CAD mutations and uridine-responsive epileptic encephalopathy. <i>Brain</i> , 2017, 140, 279-286.	3.7	106
13	The alpha-kinase family: an exceptional branch on the protein kinase tree. <i>Cellular and Molecular Life Sciences</i> , 2010, 67, 875-890.	2.4	104
14	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. <i>American Journal of Human Genetics</i> , 2018, 102, 1143-1157.	2.6	94
15	The structure-function relationship of the <i>Aspergillus fumigatus</i> cyp51A L98H conversion by site-directed mutagenesis: The mechanism of L98H azole resistance. <i>Fungal Genetics and Biology</i> , 2011, 48, 1062-1070.	0.9	92
16	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. <i>American Journal of Human Genetics</i> , 2011, 88, 608-615.	2.6	88
17	Membrane Topology and Intracellular Processing of Cyclin M2 (CNNM2). <i>Journal of Biological Chemistry</i> , 2012, 287, 13644-13655.	1.6	86
18	Germline Mutations in the Spindle Assembly Checkpoint Genes BUB1 and BUB3 Are Risk Factors for Colorectal Cancer. <i>Gastroenterology</i> , 2013, 145, 544-547.	0.6	86

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19	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. <i>Nature Genetics</i> , 2018, 50, 120-129.	9.4	86
20	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. <i>American Journal of Human Genetics</i> , 2017, 101, 478-484.	2.6	84
21	Gene structure and mutant alleles of PCDH15: nonsyndromic deafness DFNB23 and type 1 Usher syndrome. <i>Human Genetics</i> , 2008, 124, 215-223.	1.8	81
22	Whole-exome sequencing reveals <i>LRP5</i> mutations and canonical Wnt signaling associated with hepatic cystogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 5343-5348.	3.3	79
23	Microcephaly with Simplified Gyration, Epilepsy, and Infantile Diabetes Linked to Inappropriate Apoptosis of Neural Progenitors. <i>American Journal of Human Genetics</i> , 2011, 89, 265-276.	2.6	77
24	A Novel Homozygous Nonsense Mutation in <i>CABP4</i> Causes Congenital Cone-Rod Synaptic Disorder. <i>Investigative Ophthalmology and Visual Science</i> , 2009, 50, 2344.		76
25	Antisense oligonucleotide-based treatment of retinitis pigmentosa caused by <i>USH2A</i> exon 13 mutations. <i>Molecular Therapy</i> , 2021, 29, 2441-2455.	3.7	75
26	<i>SMAD2</i> Mutations Are Associated with Arterial Aneurysms and Dissections. <i>Human Mutation</i> , 2015, 36, 1145-1149.	1.1	74
27	Heterozygous Loss-of-Function Mutations in <i>DLL4</i> Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	2.6	73
28	<i>SDHA</i> mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. <i>European Journal of Human Genetics</i> , 2015, 23, 202-209.	1.4	71
29	<i>CHD3</i> helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
30	Genotype-Phenotype Correlation in <i>DFNB8/10</i> Families with <i>TMPRSS3</i> Mutations. <i>JARO - Journal of the Association for Research in Otolaryngology</i> , 2011, 12, 753-766.	0.9	69
31	Mutations in <i>PCBD1</i> Cause Hypomagnesemia and Renal Magnesium Wasting. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 574-586.	3.0	68
32	Gene polymorphisms in pattern recognition receptors and susceptibility to idiopathic recurrent vulvovaginal candidiasis. <i>Frontiers in Microbiology</i> , 2014, 5, 483.	1.5	66
33	Mutations of <i>LRTOMT</i> , a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. <i>Nature Genetics</i> , 2008, 40, 1335-1340.	9.4	65
34	Recurrent Mutations in the Basic Domain of <i>TWIST2</i> Cause Ablepharon Macrostomia and Barber-Say Syndromes. <i>American Journal of Human Genetics</i> , 2015, 97, 99-110.	2.6	61
35	De novo <i>WNT5A</i> associated autosomal dominant Robinow syndrome suggests specificity of genotype and phenotype. <i>Clinical Genetics</i> , 2015, 87, 34-41.	1.0	56
36	Allelic Mutations of <i>KITLG</i> , Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. <i>American Journal of Human Genetics</i> , 2015, 97, 647-660.	2.6	55

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37	Identification of Novel Candidate Genes for Early-Onset Colorectal Cancer Susceptibility. <i>PLoS Genetics</i> , 2016, 12, e1005880.	1.5	52
38	Homology modelling and spectroscopy, a never-ending love story. <i>European Biophysics Journal</i> , 2010, 39, 551-563.	1.2	51
39	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. <i>American Journal of Human Genetics</i> , 2010, 87, 146-153.	2.6	50
40	Functional Analysis of the Kv1.1 N255D Mutation Associated with Autosomal Dominant Hypomagnesemia. <i>Journal of Biological Chemistry</i> , 2010, 285, 171-178.	1.6	50
41	Role of the $\hat{\pm}$ -Kinase Domain in Transient Receptor Potential Melastatin 6 Channel and Regulation by Intracellular ATP. <i>Journal of Biological Chemistry</i> , 2008, 283, 19999-20007.	1.6	48
42	<i>&lt;i&gt;NPHP4&lt;/i&gt; Variants Are Associated With Pleiotropic Heart Malformations. <i>Circulation Research</i>, 2012, 110, 1564-1574.</i>	2.0	46
43	Retinal-Based Proton Pumping in the Near Infrared. <i>Journal of the American Chemical Society</i> , 2017, 139, 2338-2344.	6.6	45
44	Genotype and clinical care correlations in craniosynostosis: Findings from a cohort of 630 Australian and New Zealand patients. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2013, 163, 259-270.	0.7	44
45	A dsRNA-binding protein of a complex invertebrate DNA virus suppresses the <i>Drosophila</i> RNAi response. <i>Nucleic Acids Research</i> , 2014, 42, 12237-12248.	6.5	44
46	Identification of a novel MET mutation in high-grade glioma resulting in an auto-active intracellular protein. <i>Acta Neuropathologica</i> , 2015, 130, 131-144.	3.9	43
47	Therapeutic NOTCH3 cysteine correction in CADASIL using exon skipping: <i>&lt;i&gt;in vitro&lt;/i&gt;</i> proof of concept. <i>Brain</i> , 2016, 139, 1123-1135.	3.7	43
48	Secondary and tertiary structure modeling reveals effects of novel mutations in polycystic liver disease genes <i>&lt;i&gt;PRKCSH&lt;/i&gt;</i> and <i>&lt;i&gt;SEC63&lt;/i&gt;</i> . <i>Clinical Genetics</i> , 2010, 78, 47-56.	1.0	42
49	Mutation in subdomain G' of mitochondrial elongation factor G1 is associated with combined OXPHOS deficiency in fibroblasts but not in muscle. <i>European Journal of Human Genetics</i> , 2011, 19, 275-279.	1.4	42
50	Clinical and molecular characterizations of novel <i>&lt;i&gt;POU3F4&lt;/i&gt;</i> mutations reveal that DFN3 is due to null function of POU3F4 protein. <i>Physiological Genomics</i> , 2009, 39, 195-201.	1.0	37
51	A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. <i>Genetics in Medicine</i> , 2018, 20, 1061-1068.	1.1	37
52	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. <i>American Journal of Human Genetics</i> , 2020, 107, 164-172.	2.6	37
53	Maternal Uniparental Isodisomy of Chromosome 6 Reveals a TULP1 Mutation as a Novel Cause of Cone Dysfunction. <i>Ophthalmology</i> , 2013, 120, 1239-1246.	2.5	36
54	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. <i>American Journal of Human Genetics</i> , 2017, 101, 824-832.	2.6	36

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55	Aggregation of population-based genetic variation over protein domain homologues and its potential use in genetic diagnostics. <i>Human Mutation</i> , 2017, 38, 1454-1463.	1.1	36
56	Germline activating TYK2 mutations in pediatric patients with two primary acute lymphoblastic leukemia occurrences. <i>Leukemia</i> , 2017, 31, 821-828.	3.3	35
57	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.	2.6	35
58	Mutation in mitochondrial complex IV subunit COX5A causes pulmonary arterial hypertension, lactic acidemia, and failure to thrive. <i>Human Mutation</i> , 2017, 38, 692-703.	1.1	32
59	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. <i>Human Genetics</i> , 2018, 137, 389-400.	1.8	32
60	A Patient with Complex I Deficiency Caused by a Novel ACAD9 Mutation Not Responding to Riboflavin Treatment. <i>JIMD Reports</i> , 2013, 12, 37-45.	0.7	29
61	SNP Linkage Analysis and Whole Exome Sequencing Identify a Novel POU4F3 Mutation in Autosomal Dominant Late-Onset Nonsyndromic Hearing Loss (DFNA15). <i>PLoS ONE</i> , 2013, 8, e79063.	1.1	28
62	Whole exome sequencing identifies a heterozygous missense variant in the PRDM5 gene in a family with Axenfeld-Rieger syndrome. <i>Neurogenetics</i> , 2016, 17, 17-23.	0.7	28
63	LRP5 variants may contribute to ADPKD. <i>European Journal of Human Genetics</i> , 2016, 24, 237-242.	1.4	28
64	Bacterial CS2 Hydrolases from <i>Acidithiobacillus thiooxidans</i> Strains Are Homologous to the Archaeal Catenane CS2 Hydrolase. <i>Journal of Bacteriology</i> , 2013, 195, 4046-4056.	1.0	27
65	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2015, 96, 386-396.	2.6	27
66	A catalytic defect in mitochondrial respiratory chain complex I due to a mutation in NDUFS2 in a patient with Leigh syndrome. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 168-175.	1.8	26
67	Modulation of spectral properties and pump activity of proteorhodopsins by retinal analogues. <i>Biochemical Journal</i> , 2015, 467, 333-343.	1.7	26
68	Functional characterization of TBR1 variants in neurodevelopmental disorder. <i>Scientific Reports</i> , 2018, 8, 14279.	1.6	26
69	Mutations in GDF11 and the extracellular antagonist, Follistatin, as a likely cause of Mendelian forms of orofacial clefting in humans. <i>Human Mutation</i> , 2019, 40, 1813-1825.	1.1	26
70	Mass Spectrometry Analysis of Hecpidin Peptides in Experimental Mouse Models. <i>PLoS ONE</i> , 2011, 6, e16762.	1.1	25
71	Highly conserved nucleotide phosphatase essential for membrane lipid homeostasis in <i>Streptococcus pneumoniae</i> . <i>Molecular Microbiology</i> , 2016, 101, 12-26.	1.2	24
72	Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. <i>Human Mutation</i> , 2017, 38, 1786-1795.	1.1	24

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73	A Gate Hinge Controls the Epithelial Calcium Channel TRPV5. <i>Scientific Reports</i> , 2017, 7, 45489.	1.6	23
74	Mutations in <i>CYB561</i> Causing a Novel Orthostatic Hypotension Syndrome. <i>Circulation Research</i> , 2018, 122, 846-854.	2.0	22
75	Toward clinical and molecular understanding of pathogenic variants in the <i>ZBTB18</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 393-400.	0.6	22
76	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. <i>Genetics in Medicine</i> , 2021, 23, 1705-1714.	1.1	22
77	Mutations in <i>AGBL5</i> , Encoding $\hat{\pm}$ -Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
78	A novel COCH mutation associated with autosomal dominant nonsyndromic hearing loss disrupts the structural stability of the vWFA2 domain. <i>Journal of Molecular Medicine</i> , 2012, 90, 1321-1331.	1.7	20
79	Nonsyndromic Hearing Loss Caused by USH1G Mutations. <i>Ear and Hearing</i> , 2015, 36, 205-211.	1.0	20
80	The effect of novel mutations on the structure and enzymatic activity of unconventional myosins associated with autosomal dominant non-syndromic hearing loss. <i>Open Biology</i> , 2014, 4, 140107.	1.5	19
81	A missense variant of the ATP1A2 gene is associated with a novel phenotype of progressive sensorineural hearing loss associated with migraine. <i>European Journal of Human Genetics</i> , 2015, 23, 639-645.	1.4	18
82	A role for repressive complexes and H3K9 di-methylation in PRDM5-associated brittle cornea syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 6565-6579.	1.4	17
83	Identification of recurrent and novel mutations in TULP1 in Pakistani families with early-onset retinitis pigmentosa. <i>Molecular Vision</i> , 2012, 18, 1226-37.	1.1	17
84	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. <i>Genetics in Medicine</i> , 2020, 22, 797-802.	1.1	15
85	The moonlighting function of pyruvate carboxylase resides in the non-catalytic end of the TIM barrel. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2010, 1803, 1038-1042.	1.9	14
86	Familial hemiplegic migraine mutations affect Na,K-ATPase domain interactions. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 2173-2179.	1.8	14
87	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. <i>Genetics in Medicine</i> , 2020, 22, 1061-1068.	1.1	14
88	Submembranous recruitment of creatine kinase B supports formation of dynamic actin-based protrusions of macrophages and relies on its C-terminal flexible loop. <i>European Journal of Cell Biology</i> , 2015, 94, 114-127.	1.6	13
89	A germ line mutation in cathepsin B points toward a role in asparaginase pharmacokinetics. <i>Blood</i> , 2014, 124, 3027-3029.	0.6	12
90	Novel <i>IRF6</i> Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing. <i>Journal of Dental Research</i> , 2017, 96, 179-185.	2.5	12

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91	Identification of a novel inactivating mutation in Isocitrate Dehydrogenase 1 (IDH1-R314C) in a high grade astrocytoma. <i>Scientific Reports</i> , 2016, 6, 30486.	1.6	11
92	NewProt – a protein engineering portal. <i>Protein Engineering, Design and Selection</i> , 2017, 30, 441-447.	1.0	11
93	Novel GANAB variants associated with polycystic liver disease. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 302.	1.2	11
94	A novel (Leu183Pro-)mutation in the HFE-gene co-inherited with the Cys282Tyr mutation in two unrelated Dutch hemochromatosis patients. <i>Blood Cells, Molecules, and Diseases</i> , 2008, 40, 334-338.	0.6	10
95	Structural model of a putrescine-cadaverine permease from <i>Trypanosoma cruzi</i> predicts residues vital for transport and ligand binding. <i>Biochemical Journal</i> , 2013, 452, 423-432.	1.7	10
96	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	1.1	9
97	Identification of a de novo variant in <i>CHUK</i> in a patient with an EEC/AEC syndrome-like phenotype and hypogammaglobulinemia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1813-1820.	0.7	8
98	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 405-411.	2.6	8
99	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6419.	1.8	8
100	Variation in Genes of $\beta$ -glucan Recognition Pathway and Susceptibility to Opportunistic Infections in HIV-Positive Patients. <i>Immunological Investigations</i> , 2011, 40, 735-750.	1.0	7
101	Homozygosity mapping identifies genetic defects in four consanguineous families with retinal dystrophy from Pakistan. <i>Clinical Genetics</i> , 2013, 84, 290-293.	1.0	7
102	Deficiency of the human cysteine protease inhibitor cystatin M/E causes hypotrichosis and dry skin. <i>Genetics in Medicine</i> , 2019, 21, 1559-1567.	1.1	7
103	Novel defect in phosphatidylinositol 4-kinase type 2 $\alpha$ ( <i>PI4K2A</i> ) at the membrane-enzyme interface is associated with metabolic cutis laxa. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1382-1391.	1.7	7
104	Negative Constraints Underlie the ErbB Specificity of Epidermal Growth Factor-like Ligands. <i>Journal of Biological Chemistry</i> , 2006, 281, 40033-40040.	1.6	5
105	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2021, 23, 1246-1254.	1.1	5
106	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the <i>AGR2</i> gene. <i>Journal of Medical Genetics</i> , 2022, 59, 993-1001.	1.5	5
107	Scrutinizing pathogenicity of the USH2A c.2276G>T; p.(Cys759Phe) variant. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	5
108	Role of the C-terminal linear region of EGF-like growth factors in ErbB specificity. <i>Growth Factors</i> , 2009, 27, 163-172.	0.5	4

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109	Investigating the active site of human trimethyllysine hydroxylase. <i>Biochemical Journal</i> , 2019, 476, 1109-1119.	1.7	4
110	Status quo of annotation of human disease variants. <i>BMC Bioinformatics</i> , 2013, 14, 352.	1.2	3
111	The future of HOPE: what can and cannot be predicted about the molecular effects of a disease causing point mutation in a protein?. <i>EMBnet Journal</i> , 2011, 17, 25.	0.2	3
112	Point Mutation Approach to Reduce Antigenicity of Interferon Beta. <i>International Journal of Peptide Research and Therapeutics</i> , 2020, 26, 1353-1361.	0.9	2
113	Phosphorylation target site specificity for AGC kinases DMPK E and I $\beta$ 2. <i>Journal of Cellular Biochemistry</i> , 2012, 113, 2126-2135.	1.2	1
114	Bifunctional protein PCBD2 operates as a co-factor for hepatocyte nuclear factor 1 $\beta$ and modulates gene transcription. <i>FASEB Journal</i> , 2021, 35, e21366.	0.2	1
115	Front Cover, Volume 40, Issue 10. <i>Human Mutation</i> , 2019, 40, i.	1.1	0
116	A Germline Mutation in Cathepsin B in a Child with ALL Points towards a Key Role for This Enzyme in L-Asparaginase Pharmacokinetics. <i>Blood</i> , 2012, 120, 2458-2458.	0.6	0