

# Gerard Pals

## List of Publications by Year in descending order

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141  
papers

13,868  
citations

30070

54  
h-index

20961

115  
g-index

144  
all docs

144  
docs citations

144  
times ranked

14648  
citing authors

#	ARTICLE	IF	CITATIONS
1	Relative quantification of 40 nucleic acid sequences by multiplex ligation-dependent probe amplification. <i>Nucleic Acids Research</i> , 2002, 30, 57e-57.	14.5	2,173
2	Biallelic Inactivation of <i>BRCA2</i> in Fanconi Anemia. <i>Science</i> , 2002, 297, 606-609.	12.6	1,072
3	Long-term sequelae of <i>Helicobacter pylori</i> gastritis. <i>Lancet, The</i> , 1995, 345, 1525-1528.	13.7	720
4	Dysplastic changes in prophylactically removed Fallopian tubes of women predisposed to developing ovarian cancer. <i>Journal of Pathology</i> , 2001, 195, 451-456.	4.5	681
5	Mutations in <i>SMAD3</i> cause a syndromic form of aortic aneurysms and dissections with early-onset osteoarthritis. <i>Nature Genetics</i> , 2011, 43, 121-126.	21.4	583
6	Fanconi anemia is associated with a defect in the <i>BRCA2</i> partner <i>PALB2</i> . <i>Nature Genetics</i> , 2007, 39, 159-161.	21.4	402
7	Expression cloning of a cDNA for the major Fanconi anaemia gene, <i>FAA</i> . <i>Nature Genetics</i> , 1996, 14, 320-323.	21.4	401
8	The DNA helicase <i>BRIP1</i> is defective in Fanconi anemia complementation group J. <i>Nature Genetics</i> , 2005, 37, 934-935.	21.4	399
9	X-linked inheritance of Fanconi anemia complementation group B. <i>Nature Genetics</i> , 2004, 36, 1219-1224.	21.4	271
10	<i>PPIB</i> Mutations Cause Severe Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2009, 85, 521-527.	6.2	257
11	Large genomic deletions and duplications in the <i>BRCA1</i> gene identified by a novel quantitative method. <i>Cancer Research</i> , 2003, 63, 1449-53.	0.9	233
12	Seroconversion for <i>Helicobacter pylori</i> . <i>Lancet, The</i> , 1993, 342, 328-331.	13.7	224
13	Phenotypic spectrum of the <i>SMAD3</i> -related aneurysms-osteoarthritis syndrome. <i>Journal of Medical Genetics</i> , 2012, 49, 47-57.	3.2	221
14	Comprehensive molecular screening of the <i>FBN1</i> gene favors locus homogeneity of classical Marfan syndrome. <i>Human Mutation</i> , 2004, 24, 140-146.	2.5	210
15	Classification of Osteogenesis Imperfecta revisited. <i>European Journal of Medical Genetics</i> , 2010, 53, 1-5.	1.3	184
16	Splice-Site Mutations in the Axonemal Outer Dynein Arm Docking Complex Gene <i>CCDC114</i> Cause Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 92, 88-98.	6.2	176
17	<i>PLS3</i> Mutations in X-Linked Osteoporosis with Fractures. <i>New England Journal of Medicine</i> , 2013, 369, 1529-1536.	27.0	171
18	Mutations in <i>ZMYND10</i> , a Gene Essential for Proper Axonemal Assembly of Inner and Outer Dynein Arms in Humans and Flies, Cause Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 346-356.	6.2	167

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19	Osteogenesis Imperfecta: A Review with Clinical Examples. <i>Molecular Syndromology</i> , 2011, 2, 1-20.	0.8	159
20	Genomic deletions of MSH2 and MLH1 in colorectal cancer families detected by a novel mutation detection approach. <i>British Journal of Cancer</i> , 2002, 87, 892-897.	6.4	155
21	Beneficial Outcome of Losartan Therapy Depends on Type of <i>FBN1</i> Mutation in Marfan Syndrome. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 383-388.	5.1	143
22	A genome-wide scan for preeclampsia in the Netherlands. <i>European Journal of Human Genetics</i> , 2001, 9, 758-764.	2.8	140
23	Searching for preeclampsia genes: the current position. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2002, 105, 94-113.	1.1	138
24	Mutations in <i>PIH1D3</i> Cause X-Linked Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. <i>American Journal of Human Genetics</i> , 2017, 100, 160-168.	6.2	136
25	Mutations in <i>FKBP10</i> , which result in Bruck syndrome and recessive forms of osteogenesis imperfecta, inhibit the hydroxylation of telopeptide lysines in bone collagen. <i>Human Molecular Genetics</i> , 2013, 22, 1-17.	2.9	135
26	Localization of the Gene for Rapidly Progressive Autosomal Dominant Parkinsonism and Dementia with Pallido-Ponto-Nigral Degeneration to Chromosome 17q21. <i>Human Molecular Genetics</i> , 1996, 5, 151-154.	2.9	120
27	Genome Scan for Familial Abdominal Aortic Aneurysm Using Sex and Family History as Covariates Suggests Genetic Heterogeneity and Identifies Linkage to Chromosome 19q13. <i>Circulation</i> , 2004, 109, 2103-2108.	1.6	120
28	Mutations Near Amino End of $\alpha 1(I)$ Collagen Cause Combined Osteogenesis Imperfecta/Ehlers-Danlos Syndrome by Interference with N-propeptide Processing. <i>Journal of Biological Chemistry</i> , 2005, 280, 19259-19269.	3.4	118
29	The parent-of-origin effect of 10q22 in pre-eclamptic females coincides with two regions clustered for genes with down-regulated expression in androgenetic placentas. <i>Molecular Human Reproduction</i> , 2004, 10, 589-598.	2.8	116
30	Hypomorphic Mutations in the Gene Encoding a Key Fanconi Anemia Protein, <i>FANCD2</i> , Sustain a Significant Group of FA-D2 Patients with Severe Phenotype. <i>American Journal of Human Genetics</i> , 2007, 80, 895-910.	6.2	115
31	Genotype impacts survival in Marfan syndrome. <i>European Heart Journal</i> , 2016, 37, 3285-3290.	2.2	114
32	Familial abdominal aortic aneurysms: Collection of 233 multiplex families. <i>Journal of Vascular Surgery</i> , 2003, 37, 340-345.	1.1	110
33	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. <i>European Journal of Human Genetics</i> , 2012, 20, 11-19.	2.8	107
34	The Risk for Type B Aortic Dissection in Marfan Syndrome. <i>Journal of the American College of Cardiology</i> , 2015, 65, 246-254.	2.8	107
35	Identical twins with discordant karyotypes. <i>Prenatal Diagnosis</i> , 1999, 19, 72-76.	2.3	105
36	The Genetic Basis of Pachyonychia Congenita. <i>Journal of Investigative Dermatology Symposium Proceedings</i> , 2005, 10, 21-30.	0.8	98

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37	Combined exome and whole-genome sequencing identifies mutations in <i>ARMC4</i> as a cause of primary ciliary dyskinesia with defects in the outer dynein arm. <i>Journal of Medical Genetics</i> , 2014, 51, 61-67.	3.2	88
38	Familial Abdominal Aortic Aneurysm: a Systematic Review of a Genetic Background. <i>European Journal of Vascular and Endovascular Surgery</i> , 2002, 24, 105-116.	1.5	84
39	Generation and Molecular Characterization of Head and Neck Squamous Cell Lines of Fanconi Anemia Patients. <i>Cancer Research</i> , 2005, 65, 1271-1276.	0.9	76
40	<i>SMAD2</i> Mutations Are Associated with Arterial Aneurysms and Dissections. <i>Human Mutation</i> , 2015, 36, 1145-1149.	2.5	74
41	Relationship between fibrillin-1 genotype and severity of cardiovascular involvement in Marfan syndrome. <i>Heart</i> , 2017, 103, 1795-1799.	2.9	74
42	High-resolution mapping of molecular events associated with immortalization, transformation, and progression to breast cancer in the MCF10 model. <i>Breast Cancer Research and Treatment</i> , 2006, 96, 177-186.	2.5	73
43	Mutations in the gene for methylenetetrahydrofolate reductase, homocysteine levels, and vitamin status in women with a history of preeclampsia. <i>American Journal of Obstetrics and Gynecology</i> , 2001, 184, 394-402.	1.3	71
44	Should chromosome breakage studies be performed in patients with VACTERL association?. <i>American Journal of Medical Genetics, Part A</i> , 2005, 137A, 55-58.	1.2	69
45	Clinical utility gene card for: osteogenesis imperfecta. <i>European Journal of Human Genetics</i> , 2013, 21, 1-4.	2.8	68
46	Genetic subtyping of Fanconi anemia by comprehensive mutation screening. <i>Human Mutation</i> , 2008, 29, 159-166.	2.5	65
47	The clinical spectrum of complete FBN1 allele deletions. <i>European Journal of Human Genetics</i> , 2011, 19, 247-252.	2.8	65
48	Inflammation Aggravates Disease Severity in Marfan Syndrome Patients. <i>PLoS ONE</i> , 2012, 7, e32963.	2.5	65
49	<i>Helicobacter pylori</i> Serology in Patients with Gastric Carcinoma. <i>Scandinavian Journal of Gastroenterology</i> , 1993, 28, 433-437.	1.5	61
50	Fine-Mapping Loss of Gene Architecture at the CDKN2B (p15INK4b), CDKN2A (p14ARF, p16INK4a), and MTAP Genes in Head and Neck Squamous Cell Carcinoma. <i>JAMA Otolaryngology</i> , 2006, 132, 409.	1.2	60
51	Evaluation and application of denaturing HPLC for mutation detection in Marfan syndrome: Identification of 20 novel mutations and two novel polymorphisms in the FBN1 gene. <i>Human Mutation</i> , 2002, 19, 443-456.	2.5	58
52	The role of type III collagen in spontaneous cervical arterial dissections. <i>Annals of Neurology</i> , 1998, 43, 494-498.	5.3	57
53	Delineating Genetic Pathways of Disease Progression in Head and Neck Squamous Cell Carcinoma. <i>JAMA Otolaryngology</i> , 2003, 129, 702.	1.2	57
54	BRCA1 and BRCA2 germline mutation analysis in the Indonesian population. <i>Breast Cancer Research and Treatment</i> , 2007, 106, 297-304.	2.5	56

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55	Fanconi Anemia. JAMA Otolaryngology, 2005, 131, 635.	1.2	55
56	Exhaled molecular profiles in the assessment of cystic fibrosis and primary ciliary dyskinesia. Journal of Cystic Fibrosis, 2013, 12, 454-460.	0.7	55
57	Effect of single and repeated doses of oral omeprazole on gastric acid and pepsin secretion and fasting serum gastrin and serum pepsinogen I levels. Digestive Diseases and Sciences, 1986, 31, 561-566.	2.3	53
58	Genome-Wide Linkage in a Large Dutch Consanguineous Family Maps a Locus for Intracranial Aneurysms to Chromosome 2p13. Stroke, 2004, 35, 2276-2281.	2.0	47
59	The many faces of aggressive aortic pathology: Loeys-Dietz syndrome. Netherlands Heart Journal, 2008, 16, 299-304.	0.8	47
60	Familial Ehlers-Danlos syndrome with lethal arterial events caused by a mutation in <i>COL5A1</i> . American Journal of Medical Genetics, Part A, 2015, 167, 1196-1203.	1.2	47
61	Gastric Non-Hodgkin Lymphomas of Mucosa-Associated Lymphoid Tissue Are not Associated With More Aggressive <i>Helicobacter pylori</i> Strains as Identified by CagA. American Journal of Clinical Pathology, 1996, 106, 670-675.	0.7	46
62	Genetics of urinary pepsinogen: A new hypothesis. Human Genetics, 1984, 65, 385-390.	3.8	45
63	CRTAP mutations in lethal and severe osteogenesis imperfecta: the importance of combining biochemical and molecular genetic analysis. European Journal of Human Genetics, 2009, 17, 1560-1569.	2.8	44
64	Clinical and genetic evaluation of thirty ovarian cancer families. American Journal of Obstetrics and Gynecology, 1998, 178, 85-90.	1.3	43
65	Collagen transport and related pathways in Osteogenesis Imperfecta. Human Genetics, 2021, 140, 1121-1141.	3.8	42
66	Ehlers-Danlos arthrochalasia type (VIIA-B) - expanding the phenotype: from prenatal life through adulthood. Clinical Genetics, 2012, 82, 121-130.	2.0	40
67	Human pepsinogen C (progastricsin) polymorphism: Evidence for a single locus located at 6p21.1-pter. Genomics, 1989, 4, 137-145.	2.9	39
68	Expression of differentiation and proliferation related proteins in epithelium of prophylactically removed ovaries from women with a hereditary female adnexal cancer predisposition. Histopathology, 2003, 43, 26-32.	2.9	38
69	Genome-wide Linkage in Three Dutch Families Maps a Locus for Abdominal Aortic Aneurysms to Chromosome 19q13.3. European Journal of Vascular and Endovascular Surgery, 2005, 30, 29-35.	1.5	38
70	Inhibition of TGF $\beta$ 2 signaling decreases osteogenic differentiation of fibrodysplasia ossificans progressiva fibroblasts in a novel in vitro model of the disease. Bone, 2016, 84, 169-180.	2.9	38
71	Diagnostic yield of a targeted gene panel in primary ciliary dyskinesia patients. Human Mutation, 2018, 39, 653-665.	2.5	38
72	Type III Collagen Deficiency in Saccular Intracranial Aneurysms. Stroke, 1999, 30, 1628-1631.	2.0	36

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73	Offering Preconceptional Cystic Fibrosis Carrier Couple Screening in the Absence of Established Preconceptional Care Services. <i>Public Health Genomics</i> , 2003, 6, 5-13.	1.0	36
74	Incomplete segregation of MYH11 variants with thoracic aortic aneurysms and dissections and patent ductus arteriosus. <i>European Journal of Human Genetics</i> , 2013, 21, 487-493.	2.8	36
75	Complete COL1A1 allele deletions in osteogenesis imperfecta. <i>Genetics in Medicine</i> , 2010, 12, 736-741.	2.4	35
76	Interaction between KDELR2 and HSP47 as a Key Determinant in Osteogenesis Imperfecta Caused by Bi-allelic Variants in KDELR2. <i>American Journal of Human Genetics</i> , 2020, 107, 989-999.	6.2	35
77	Effect of high dose omeprazole on gastric pepsin secretion and serum pepsinogen levels in man. <i>European Journal of Clinical Pharmacology</i> , 1988, 35, 173-176.	1.9	34
78	LINKAGE AND ASSOCIATION STUDIES OF IL1B AND IL1RN GENE POLYMORPHISMS IN PREECLAMPSIA. <i>Hypertension in Pregnancy</i> , 2002, 21, 23-38.	1.1	34
79	The revised role of TGF- $\beta$ 2 in aortic aneurysms in Marfan syndrome. <i>Netherlands Heart Journal</i> , 2015, 23, 116-121.	0.8	34
80	Neonatal Marfan syndrome: clinical report and review of the literature. <i>Clinical Dysmorphology</i> , 2005, 14, 81-84.	0.3	32
81	Homozygosity for a FBN1 missense mutation: clinical and molecular evidence for recessive Marfan syndrome. <i>European Journal of Human Genetics</i> , 2007, 15, 930-935.	2.8	32
82	Relation between genotype and left-ventricular dilatation in patients with Marfan syndrome. <i>Gene</i> , 2014, 534, 40-43.	2.2	32
83	[18F]NaF PET/CT scan as an early marker of heterotopic ossification in fibrodysplasia ossificans progressiva. <i>Bone</i> , 2018, 109, 143-146.	2.9	31
84	Variation in gene copy number and polymorphism of the human salivary amylase isoenzyme system in Caucasians. <i>Human Genetics</i> , 1992, 89, 213-22.	3.8	28
85	Polymorphisms in the tumor necrosis factor and lymphotoxin- $\alpha$ gene region and preeclampsia. <i>Obstetrics and Gynecology</i> , 2001, 98, 612-619.	2.4	28
86	Compound-heterozygous Marfan syndrome. <i>European Journal of Medical Genetics</i> , 2009, 52, 1-5.	1.3	28
87	Periodontal ligament fibroblasts as a cell model to study osteogenesis and osteoclastogenesis in fibrodysplasia ossificans progressiva. <i>Bone</i> , 2018, 109, 168-177.	2.9	28
88	Ehlers-Danlos syndrome type IV: unusual congenital anomalies in a mother and son with a COL3A1 mutation and a normal collagen III protein profile. <i>Clinical Genetics</i> , 2003, 63, 224-227.	2.0	26
89	A rapid and sensitive approach to mutation detection using real-time polymerase chain reaction and melting curve analyses, using BRCA1 as an example*. <i>Molecular Diagnosis and Therapy</i> , 1999, 4, 241-246.	1.1	25
90	The clinical spectrum of missense mutations of the first aspartic acid of cbEGF-like domains in fibrillin-1 including a recessive family. <i>Human Mutation</i> , 2010, 31, E1915-E1927.	2.5	25

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91	Renal handling of pepsinogens A and C in man. <i>Clinical Science</i> , 1988, 75, 649-654.	4.3	23
92	Determinants of Fasting and Post-Methionine Homocysteine Levels in Families Predisposed to Hyperhomocysteinemia and Premature Vascular Disease. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 1999, 19, 1316-1324.	2.4	23
93	Lethal/Severe Osteogenesis Imperfecta in a Large Family: A Novel Homozygous LEPRE1 Mutation and Bone Histological Findings. <i>Pediatric and Developmental Pathology</i> , 2011, 14, 228-234.	1.0	23
94	Flare-up After Maxillofacial Surgery in a Patient With Fibrodysplasia Ossificans Progressiva: An [ <sup>18</sup> F]NaF PET/CT Study and a Systematic Review. <i>JBMR Plus</i> , 2018, 2, 55-58.	2.7	21
95	TGFB1 gene polymorphisms and inflammatory bowel disease. <i>Immunogenetics</i> , 2000, 51, 869-872.	2.4	20
96	Polymorphisms in the Tumor Necrosis Factor and Lymphotoxin- $\beta$ Gene Region and Preeclampsia. <i>Obstetrics and Gynecology</i> , 2001, 98, 612-619.	2.4	20
97	Evolution of heterotopic bone in fibrodysplasia ossificans progressiva: An [ <sup>18</sup> F]NaF PET/CT study. <i>Bone</i> , 2019, 124, 1-6.	2.9	20
98	Diagnosis and genetics of Marfan syndrome. <i>Expert Opinion on Orphan Drugs</i> , 2014, 2, 1049-1062.	0.8	18
99	Clinical Utility Gene Card for: Fibrodysplasia ossificans progressiva. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	2.8	18
100	Transdifferentiation of Human Dermal Fibroblasts to Smooth Muscle-Like Cells to Study the Effect of MYH11 and ACTA2 Mutations in Aortic Aneurysms. <i>Human Mutation</i> , 2017, 38, 439-450.	2.5	18
101	Single base mutations can be unequivocally and rapidly detected by analysis of DNA heteroduplexes, obtained with deletion-mutant instead of wild-type DNA. <i>Nucleic Acids Research</i> , 1992, 20, 6745-6746.	14.5	17
102	Detection of a single base substitution in a single cell using the LightCycler <sup>®</sup> . <i>Journal of Proteomics</i> , 2001, 47, 121-129.	2.4	16
103	Fibrodysplasia Ossificans Progressiva: What Have We Achieved and Where Are We Now? Follow-up to the 2015 Lorentz Workshop. <i>Frontiers in Endocrinology</i> , 2021, 12, 732728.	3.5	15
104	Gastric chief cell-specific transcription of the pepsinogen A gene. <i>FEBS Journal</i> , 1993, 213, 1283-1296.	0.2	14
105	Muscle weakness as presenting symptom of osteogenesis imperfecta. <i>European Journal of Pediatrics</i> , 2006, 165, 392-394.	2.7	14
106	Validation of a quantitative PCR-high-resolution melting protocol for simultaneous screening of COL1A1 and COL1A2 point mutations and large rearrangements: Application for diagnosis of osteogenesis imperfecta. <i>Human Mutation</i> , 2012, 33, 1697-1707.	2.5	14
107	RFLP for the human pepsinogen C gene (PGC). <i>Nucleic Acids Research</i> , 1988, 16, 9372-9372.	14.5	13
108	Marfan syndrome: Progress report. <i>Progress in Pediatric Cardiology</i> , 2012, 34, 9-14.	0.4	13

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109	Enzyme-linked immunosorbent assay and radioimmunoassay of serum pepsinogen A. Scandinavian Journal of Clinical and Laboratory Investigation, 1987, 47, 29-33.	1.2	12
110	Betaglycan (TGFB3) up-regulation correlates with increased TGF- $\beta$ 2 signaling in Marfan patient fibroblasts in vitro. Cardiovascular Pathology, 2018, 32, 44-49.	1.6	11
111	The first family with adult osteogenesis imperfecta caused by a novel homozygous mutation in <i>CREB3L1</i> . Molecular Genetics & Genomic Medicine, 2019, 7, e823.	1.2	11
112	The influence of omeprazole on the synthesis and secretion of pepsinogen in isolated rabbit gastric glands. Biochemical Pharmacology, 1985, 34, 3693-3699.	4.4	10
113	Type III Collagen Deficiency in a Family with Intracranial Aneurysms. Cerebrovascular Diseases, 2001, 11, 92-94.	1.7	10
114	Novel inactivating mutations of FANCC in Brazilian patients with Fanconi anemia. Human Mutation, 2006, 27, 214-214.	2.5	10
115	Establishing a molecular continuum in breast cancer DNA microarrays and benign breast disease. Cytometry, 2002, 47, 56-59.	1.8	9
116	An in vitro method to keep human aortic tissue sections functionally and structurally intact. Scientific Reports, 2018, 8, 8094.	3.3	9
117	Mechanical stress regulates bone regulatory gene expression independent of estrogen and vitamin D deficiency in rats. Journal of Orthopaedic Research, 2021, 39, 42-52.	2.3	9
118	Bioactivity of compounds secreted by symbiont bacteria of Nudibranchs from Indonesia. PeerJ, 2020, 8, e8093.	2.0	9
119	Influence of RP 40749 on basal and meal-stimulated serum-gastrin, serum-pepsinogen I, and gastrin-content of the antral mucosa in duodenal ulcer patients. Digestive Diseases and Sciences, 1985, 30, 617-623.	2.3	8
120	Discrepancies between gastric mucosal and urinary pepsinogen A patterns and in vitro synthesis and secretion of human pepsinogen. Digestive Diseases and Sciences, 1988, 33, 135-143.	2.3	8
121	Genetics and Epidemiology May Contribute to Understanding the Pathogenesis of IBD - A New Approach is Now Indicated. Canadian Journal of Gastroenterology & Hepatology, 1993, 7, 71-75.	1.7	8
122	RNA Sequencing of Creatine Transporter (SLC6A8) Deficient Fibroblasts Reveals Impairment of the Extracellular Matrix. Human Mutation, 2014, 35, 1128-1135.	2.5	8
123	Human pepsinogen A isozymogen patterns in serum and gastric mucosa. Gastroenterology, 1990, 99, 1576-1580.	1.3	7
124	Diagnostic Value of Magnetic Resonance Imaging in Fibrodysplasia Ossificans Progressiva. JBMR Plus, 2020, 4, e10363.	2.7	7
125	Prevalence and Hospital Admissions in Patients With Osteogenesis Imperfecta in The Netherlands: A Nationwide Registry Study. Frontiers in Endocrinology, 2022, 13, 869604.	3.5	7
126	Exon 6 skipping in the fanconi anemia C gene associated with a nonsense/missense mutation (775C>T) in exon 5: The first example of a nonsense mutation in one exon causing skipping of another downstream. Human Mutation, 1998, 11, S25-S27.	2.5	6



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127	A New BRCA1 Mutation in a Filipino Woman With a Family History of Breast and Ovarian Cancer. Diagnostic Molecular Pathology, 1998, 7, 164-167.	2.1	5
128	Primary ciliary dyskinesia in Volendam: Diagnostic and phenotypic features in patients with a <scp><i>CCDC114</i></scp> mutation. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 89-101.	1.6	5
129	Osteogenesis Imperfecta, Normal Collagen Folding, and Lack of Cyclophilin B. New England Journal of Medicine, 2010, 362, 1940-1942.	27.0	4
130	High-performance liquid chromatography: purification and chromatographic behaviour of molecular variants of pepsinogen A from human urine. Biomedical Applications, 1991, 571, 47-59.	1.7	3
131	Primary ciliary dyskinesia: From diagnosis to molecular mechanisms. Journal of Pediatric Genetics, 2015, 03, 115-127.	0.7	3
132	Pathogenic effect of a <i>TGFB1</i> mutation in a family with Loey's "Dietz syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00943.	1.2	3
133	Collaboration Around Rare Bone Diseases Leads to the Unique Organizational Incentive of the Amsterdam Bone Center. Frontiers in Endocrinology, 2020, 11, 481.	3.5	3
134	THIRD NORDIC CONFERENCE Pepsinogen and gastric cancer. Clinical Genetics, 1984, 26, 240-241.	2.0	2
135	Intracranial Hypertension in 2 Children With Marfan Syndrome. Journal of Child Neurology, 2008, 23, 954-955.	1.4	2
136	An in vitro model to evaluate the properties of matrices produced by fibroblasts from osteogenesis imperfecta and Ehlers-Danlos Syndrome patients. Biochemical and Biophysical Research Communications, 2020, 521, 310-317.	2.1	2
137	Phenotypic Variation in Vietnamese Osteogenesis Imperfecta Patients Sharing a Recessive P3H1 Pathogenic Variant. Genes, 2022, 13, 407.	2.4	2
138	Immunohistochemical localization of pepsinogen A and C containing cells in Barrett's oesophagus. Virchows Archiv A, Pathological Anatomy and Histopathology, 1988, 413, 11-16.	1.4	1
139	Consequences of Intramolecular Ionic Interactions for the Activation Rate of Human Pepsinogens A and C as Revealed by Molecular Modelling. Advances in Experimental Medicine and Biology, 1991, 306, 101-105.	1.6	1
140	Detection of a Single Base Substitution in Single Cells by Melting Peak Analysis Using Dual-Color Hybridization Probes. , 2002, , 77-84.		0
141	Fanconi Anemia. , 2004, , 447-451.		0