Gerard Pals

List of Publications by Year in Descending Order

139 papers	11,651 citations	51 h-index	107 g-index
144 ext. papers	12,777 ext. citations	7.7 avg, IF	5.32 L-index

#	Paper	IF	Citations
139	Prevalence and Hospital Admissions in Patients With Osteogenesis Imperfecta in The Netherlands: A Nationwide Registry Study.. *Frontiers in Endocrinology*, **2022**, 13, 869604	5.7	1
138	Fibrodysplasia Ossificans Progressiva: What Have We Achieved and Where Are We Now? Follow-up to the 2015 Lorentz Workshop. *Frontiers in Endocrinology*, **2021**, 12, 732728	5.7	1
137	Collagen transport and related pathways in Osteogenesis Imperfecta. *Human Genetics*, **2021**, 140, 1121-1141	6.1	5
136	Mechanical stress regulates bone regulatory gene expression independent of estrogen and vitamin D deficiency in rats. *Journal of Orthopaedic Research*, **2021**, 39, 42-52	3.8	4
135	Diagnostic Value of Magnetic Resonance Imaging in Fibrodysplasia Ossificans Progressiva. *JBMR Plus*, **2020**, 4, e10363	3.9	3
134	Bioactivity of compounds secreted by symbiont bacteria of Nudibranchs from Indonesia. *PeerJ*, **2020**, 8, e8093	3.1	4
133	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	3.4	0
132	Interaction between KDELR2 and HSP47 as a Key Determinant in Osteogenesis Imperfecta Caused by Bi-allelic Variants in KDELR2. *American Journal of Human Genetics*, **2020**, 107, 989-999	11	11
131	Collaboration Around Rare Bone Diseases Leads to the Unique Organizational Incentive of the Amsterdam Bone Center. *Frontiers in Endocrinology*, **2020**, 11, 481	5.7	2
130	The first family with adult osteogenesis imperfecta caused by a novel homozygous mutation in CREB3L1. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e823	2.3	8
129	Evolution of heterotopic bone in fibrodysplasia ossificans progressiva: An [F]NaF PET/CT study. *Bone*, **2019**, 124, 1-6	4.7	15
128	Pathogenic effect of a TGFBR1 mutation in a family with Loeys-Dietz syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00943	2.3	1
127	Diagnostic yield of a targeted gene panel in primary ciliary dyskinesia patients. *Human Mutation*, **2018**, 39, 653-665	4.7	23
126	Periodontal ligament fibroblasts as a cell model to study osteogenesis and osteoclastogenesis in fibrodysplasia ossificans progressiva. *Bone*, **2018**, 109, 168-177	4.7	21
125	[18F]NaF PET/CT scan as an early marker of heterotopic ossification in fibrodysplasia ossificans progressiva. *Bone*, **2018**, 109, 143-146	4.7	22
124	Betaglycan (TGFBR3) up-regulation correlates with increased TGF- signaling in Marfan patient fibroblasts in vitro. *Cardiovascular Pathology*, **2018**, 32, 44-49	3.8	6
123	Flare-Up After Maxillofacial Surgery in a Patient With Fibrodysplasia Ossificans Progressiva: An [F]-NaF PET/CT Study and a Systematic Review. *JBMR Plus*, **2018**, 2, 55-58	3.9	15

122	An in vitro method to keep human aortic tissue sections functionally and structurally intact. *Scientific Reports*, **2018**, 8, 8094	4.9	4
121	Mutations in PIH1D3 Cause X-Linked Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. *American Journal of Human Genetics*, **2017**, 100, 160-168	11	92
120	Transdifferentiation of Human Dermal Fibroblasts to Smooth Muscle-Like Cells to Study the Effect of MYH11 and ACTA2 Mutations in Aortic Aneurysms. *Human Mutation*, **2017**, 38, 439-450	4.7	12
119	Relationship between fibrillin-1 genotype and severity of cardiovascular involvement in Marfan syndrome. *Heart*, **2017**, 103, 1795-1799	5.1	49
118	Genotype impacts survival in Marfan syndrome. *European Heart Journal*, **2016**, 37, 3285-3290	9.5	78
117	Inhibition of TGF signaling decreases osteogenic differentiation of fibrodysplasia ossificans progressiva fibroblasts in a novel in vitro model of the disease. *Bone*, **2016**, 84, 169-180	4.7	32
116	The risk for type B aortic dissection in Marfan syndrome. *Journal of the American College of Cardiology*, **2015**, 65, 246-54	15.1	73
115	Beneficial Outcome of Losartan Therapy Depends on Type of FBN1 Mutation in Marfan Syndrome. *Circulation: Cardiovascular Genetics*, **2015**, 8, 383-8		107
114	Clinical Utility Gene Card for: Fibrodysplasia ossificans progressiva. *European Journal of Human Genetics*, **2015**, 23,	5.3	12
113	Familial Ehlers-Danlos syndrome with lethal arterial events caused by a mutation in COL5A1. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1196-203	2.5	39
112	SMAD2 Mutations Are Associated with Arterial Aneurysms and Dissections. *Human Mutation*, **2015**, 36, 1145-9	4.7	65
111	The revised role of TGF- in aortic aneurysms in Marfan syndrome. *Netherlands Heart Journal*, **2015**, 23, 116-21	2.2	30
110	Diagnosis and genetics of Marfan syndrome. *Expert Opinion on Orphan Drugs*, **2014**, 2, 1049-1062	1.1	16
109	Relation between genotype and left-ventricular dilatation in patients with Marfan syndrome. *Gene*, **2014**, 534, 40-3	3.8	25
108	Primary ciliary dyskinesia: From diagnosis to molecular mechanisms. *Journal of Pediatric Genetics*, **2014**, 3, 115-27	0.7	3
107	Combined exome and whole-genome sequencing identifies mutations in ARMC4 as a cause of primary ciliary dyskinesia with defects in the outer dynein arm. *Journal of Medical Genetics*, **2014**, 51, 61-7	5.8	64
106	RNA sequencing of creatine transporter (SLC6A8) deficient fibroblasts reveals impairment of the extracellular matrix. *Human Mutation*, **2014**, 35, 1128-35	4.7	6
105	PLS3 mutations in X-linked osteoporosis with fractures. *New England Journal of Medicine*, **2013**, 369, 1529-36	59.2	140

104	Splice-site mutations in the axonemal outer dynein arm docking complex gene CCDC114 cause primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 92, 88-98	11	132
103	Mutations in ZMYND10, a gene essential for proper axonemal assembly of inner and outer dynein arms in humans and flies, cause primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 93, 346-56	11	126
102	Exhaled molecular profiles in the assessment of cystic fibrosis and primary ciliary dyskinesia. *Journal of Cystic Fibrosis*, **2013**, 12, 454-60	4.1	40
101	Incomplete segregation of MYH11 variants with thoracic aortic aneurysms and dissections and patent ductus arteriosus. *European Journal of Human Genetics*, **2013**, 21, 487-93	5.3	32
100	Clinical utility gene card for: osteogenesis imperfecta. *European Journal of Human Genetics*, **2013**, 21,	5.3	41
99	Mutations in FKBP10, which result in Bruck syndrome and recessive forms of osteogenesis imperfecta, inhibit the hydroxylation of telopeptide lysines in bone collagen. *Human Molecular Genetics*, **2013**, 22, 1-17	5.6	117
98	Ehlers-Danlos arthrochalasia type (VIIA-B)--expanding the phenotype: from prenatal life through adulthood. *Clinical Genetics*, **2012**, 82, 121-30	4	31
97	Marfan syndrome: Progress report. *Progress in Pediatric Cardiology*, **2012**, 34, 9-14	0.4	13
96	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. *Human Mutation*, **2012**, 33, 1697-707	4.7	9
95	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. *European Journal of Human Genetics*, **2012**, 20, 11-9	5.3	99
94	Phenotypic spectrum of the SMAD3-related aneurysms-osteoarthritis syndrome. *Journal of Medical Genetics*, **2012**, 49, 47-57	5.8	189
93	Inflammation aggravates disease severity in Marfan syndrome patients. *PLoS ONE*, **2012**, 7, e32963	3.7	50
92	Mutations in SMAD3 cause a syndromic form of aortic aneurysms and dissections with early-onset osteoarthritis. *Nature Genetics*, **2011**, 43, 121-6	36.3	496
91	The clinical spectrum of complete FBN1 allele deletions. *European Journal of Human Genetics*, **2011**, 19, 247-52	5.3	54
90	Osteogenesis Imperfecta: A Review with Clinical Examples. *Molecular Syndromology*, **2011**, 2, 1-20	1.5	114
89	Lethal/severe osteogenesis imperfecta in a large family: a novel homozygous LEPRE1 mutation and bone histological findings. *Pediatric and Developmental Pathology*, **2011**, 14, 228-34	2.2	20
88	Osteogenesis imperfecta, normal collagen folding, and lack of cyclophilin B. *New England Journal of Medicine*, **2010**, 362, 1940-1; author reply 1941-2	59.2	4
87	Complete COL1A1 allele deletions in osteogenesis imperfecta. *Genetics in Medicine*, **2010**, 12, 736-41	8.1	31

86	Classification of Osteogenesis Imperfecta revisited. *European Journal of Medical Genetics*, **2010**, 53, 1-5	2.6	150
85	The clinical spectrum of missense mutations of the first aspartic acid of cbEGF-like domains in fibrillin-1 including a recessive family. *Human Mutation*, **2010**, 31, E1915-27	4.7	19
84	CRTAP mutations in lethal and severe osteogenesis imperfecta: the importance of combining biochemical and molecular genetic analysis. *European Journal of Human Genetics*, **2009**, 17, 1560-9	5.3	37
83	PPIB mutations cause severe osteogenesis imperfecta. *American Journal of Human Genetics*, **2009**, 85, 521-7	11	220
82	Compound-heterozygous Marfan syndrome. *European Journal of Medical Genetics*, **2009**, 52, 1-5	2.6	23
81	THIRD NORDIC CONFERENCE Pepsinogen and gastric cancer. *Clinical Genetics*, **2008**, 26, 240-241	4	
80	Intracranial hypertension in 2 children with marfan syndrome. *Journal of Child Neurology*, **2008**, 23, 954-5	2.5	2
79	Genetic subtyping of Fanconi anemia by comprehensive mutation screening. *Human Mutation*, **2008**, 29, 159-66	4.7	56
78	The many faces of aggressive aortic pathology: Loeys-Dietz syndrome. *Netherlands Heart Journal*, **2008**, 16, 299-304	2.2	40
77	Fanconi anemia is associated with a defect in the BRCA2 partner PALB2. *Nature Genetics*, **2007**, 39, 159-61	36.3	348
76	Homozygosity for a FBN1 missense mutation: clinical and molecular evidence for recessive Marfan syndrome. *European Journal of Human Genetics*, **2007**, 15, 930-5	5.3	26
75	BRCA1 and BRCA2 germline mutation analysis in the Indonesian population. *Breast Cancer Research and Treatment*, **2007**, 106, 297-304	4.4	25
74	Hypomorphic mutations in the gene encoding a key Fanconi anemia protein, FANCD2, sustain a significant group of FA-D2 patients with severe phenotype. *American Journal of Human Genetics*, **2007**, 80, 895-910	11	92
73	Novel inactivating mutations of FANCC in Brazilian patients with Fanconi anemia. *Human Mutation*, **2006**, 27, 214	4.7	4
72	Fine-mapping loss of gene architecture at the CDKN2B (p15INK4b), CDKN2A (p14ARF, p16INK4a), and MTAP genes in head and neck squamous cell carcinoma. *JAMA Otolaryngology*, **2006**, 132, 409-15		49
71	High-resolution mapping of molecular events associated with immortalization, transformation, and progression to breast cancer in the MCF10 model. *Breast Cancer Research and Treatment*, **2006**, 96, 177-86	4.4	67
70	Muscle weakness as presenting symptom of osteogenesis imperfecta. *European Journal of Pediatrics*, **2006**, 165, 392-4	4.1	13
69	Neonatal Marfan syndrome: clinical report and review of the literature. *Clinical Dysmorphology*, **2005**, 14, 81-84	0.9	25

68	The DNA helicase BRIP1 is defective in Fanconi anemia complementation group J. *Nature Genetics*, **2005**, 37, 934-5	36.3	358
67	The genetic basis of pachyonychia congenita. *Journal of Investigative Dermatology Symposium Proceedings*, **2005**, 10, 21-30	1.1	79
66	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	2.3	35
65	Should chromosome breakage studies be performed in patients with VACTERL association?. *American Journal of Medical Genetics, Part A*, **2005**, 137, 55-8	2.5	62
64	Generation and molecular characterization of head and neck squamous cell lines of fanconi anemia patients. *Cancer Research*, **2005**, 65, 1271-6	10.1	62
63	Mutations near amino end of alpha1(I) collagen cause combined osteogenesis imperfecta/Ehlers-Danlos syndrome by interference with N-propeptide processing. *Journal of Biological Chemistry*, **2005**, 280, 19259-69	5.4	98
62	Fanconi anemia: adult head and neck cancer and hematopoietic mosaicism. *JAMA Otolaryngology*, **2005**, 131, 635-9		51
61	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. *Molecular Human Reproduction*, **2004**, 10, 589-98	4.4	104
60	Genome-wide linkage in a large Dutch consanguineous family maps a locus for intracranial aneurysms to chromosome 2p13. *Stroke*, **2004**, 35, 2276-81	6.7	43
59	Genome scan for familial abdominal aortic aneurysm using sex and family history as covariates suggests genetic heterogeneity and identifies linkage to chromosome 19q13. *Circulation*, **2004**, 109, 2103-8	16.7	111
58	X-linked inheritance of Fanconi anemia complementation group B. *Nature Genetics*, **2004**, 36, 1219-24	36.3	238
57	Comprehensive molecular screening of the FBN1 gene favors locus homogeneity of classical Marfan syndrome. *Human Mutation*, **2004**, 24, 140-6	4.7	177
56	Fanconi Anemia **2004**, 447-451		
55	Offering preconceptional cystic fibrosis carrier couple screening in the absence of established preconceptional care services. *Public Health Genomics*, **2003**, 6, 5-13	1.9	33
54	Ehlers-Danlos syndrome type IV: unusual congenital anomalies in a mother and son with a COL3A1 mutation and a normal collagen III protein profile. *Clinical Genetics*, **2003**, 63, 224-7	4	20
53	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	7.3	31
52	Familial abdominal aortic aneurysms: collection of 233 multiplex families. *Journal of Vascular Surgery*, **2003**, 37, 340-5	3.5	90
51	Delineating genetic pathways of disease progression in head and neck squamous cell carcinoma. *JAMA Otolaryngology*, **2003**, 129, 702-8		53

50	Large genomic deletions and duplications in the BRCA1 gene identified by a novel quantitative method. *Cancer Research*, **2003**, 63, 1449-53	10.1	218
49	Familial abdominal aortic aneurysm: a systematic review of a genetic background. *European Journal of Vascular and Endovascular Surgery*, **2002**, 24, 105-16	2.3	67
48	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. *Human Mutation*, **2002**, 19, 443-56	4.7	51
47	Establishing a molecular continuum in breast cancer DNA microarrays and benign breast disease. *Cytometry*, **2002**, 47, 56-9		8
46	Biallelic inactivation of BRCA2 in Fanconi anemia. *Science*, **2002**, 297, 606-9	33.3	947
45	Relative quantification of 40 nucleic acid sequences by multiplex ligation-dependent probe amplification. *Nucleic Acids Research*, **2002**, 30, e57	20.1	1879
44	Linkage and association studies of IL1B and IL1RN gene polymorphisms in preeclampsia. *Hypertension in Pregnancy*, **2002**, 21, 23-38	2	31
43	Genomic deletions of MSH2 and MLH1 in colorectal cancer families detected by a novel mutation detection approach. *British Journal of Cancer*, **2002**, 87, 892-7	8.7	144
42	Searching for preeclampsia genes: the current position. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2002**, 105, 94-113	2.4	119
41	Detection of a Single Base Substitution in Single Cells by Melting Peak Analysis Using Dual-Color Hybridization Probes **2002**, 77-84		
40	Dysplastic changes in prophylactically removed Fallopian tubes of women predisposed to developing ovarian cancer. *Journal of Pathology*, **2001**, 195, 451-6	9.4	539
39	A genome-wide scan for preeclampsia in the Netherlands. *European Journal of Human Genetics*, **2001**, 9, 758-64	5.3	121
38	Mutations in the gene for methylenetetrahydrofolate reductase, homocysteine levels, and vitamin status in women with a history of preeclampsia. *American Journal of Obstetrics and Gynecology*, **2001**, 184, 394-402	6.4	66
37	Detection of a single base substitution in a single cell using the LightCycler. *Journal of Proteomics*, **2001**, 47, 121-9		15
36	Polymorphisms in the tumor necrosis factor and lymphotoxin-alpha gene region and preeclampsia. *Obstetrics and Gynecology*, **2001**, 98, 612-9	4.9	19
35	Type III collagen deficiency in a family with intracranial aneurysms. *Cerebrovascular Diseases*, **2001**, 11, 92-4	3.2	10
34	Polymorphisms in the Tumor Necrosis Factor and Lymphotoxin-Gene Region and Preeclampsia. *Obstetrics and Gynecology*, **2001**, 98, 612-619	4.9	10
33	TGFB1 gene polymorphisms and inflammatory bowel disease. *Immunogenetics*, **2000**, 51, 869-72	3.2	18

32	Identical twins with discordant karyotypes. *Prenatal Diagnosis*, **1999**, 19, 72-6	3.2	70
31	A rapid and sensitive approach to mutation detection using real-time polymerase chain reaction and melting curve analyses, using BRCA1 as an example. *Molecular Diagnosis and Therapy*, **1999**, 4, 241-6		23
30	Determinants of fasting and post-methionine homocysteine levels in families predisposed to hyperhomocysteinemia and premature vascular disease. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **1999**, 19, 1316-24	9.4	21
29	Type III collagen deficiency in saccular intracranial aneurysms. Defect in gene regulation?. *Stroke*, **1999**, 30, 1628-31	6.7	32
28	Clinical and genetic evaluation of thirty ovarian cancer families. *American Journal of Obstetrics and Gynecology*, **1998**, 178, 85-90	6.4	40
27	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**, Suppl 1, S25-7	4.7	5
26	The role of type III collagen in spontaneous cervical arterial dissections. *Annals of Neurology*, **1998**, 43, 494-8	9.4	51
25	A new BRCA1 mutation in a Filipino woman with a family history of breast and ovarian cancer. *Diagnostic Molecular Pathology*, **1998**, 7, 164-7		5
24	Gastric non-Hodgkin lymphomas of mucosa-associated lymphoid tissue are not associated with more aggressive Helicobacter pylori strains as identified by CagA. *American Journal of Clinical Pathology*, **1996**, 106, 670-5	1.9	40
23	Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. *Nature Genetics*, **1996**, 14, 320-3	36.3	376
22	Localization of the gene for rapidly progressive autosomal dominant parkinsonism and dementia with pallido-ponto-nigral degeneration to chromosome 17q21. *Human Molecular Genetics*, **1996**, 5, 151-4	5.6	114
21	Long-term sequelae of Helicobacter pylori gastritis. *Lancet, The*, **1995**, 345, 1525-8	40	632
20	Seroconversion for Helicobacter pylori. *Lancet, The*, **1993**, 342, 328-31	40	192
19	Helicobacter pylori serology in patients with gastric carcinoma. *Scandinavian Journal of Gastroenterology*, **1993**, 28, 433-7	2.4	52
18	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		8
17	Gastric chief cell-specific transcription of the pepsinogen A gene. *FEBS Journal*, **1993**, 213, 1283-96		12
16	Single base mutations can be unequivocally and rapidly detected by analysis of DNA heteroduplexes, obtained with deletion-mutant instead of wild-type DNA. *Nucleic Acids Research*, **1992**, 20, 6745-6	20.1	16
15	Variation in gene copy number and polymorphism of the human salivary amylase isoenzyme system in Caucasians. *Human Genetics*, **1992**, 89, 213-22	6.3	26

14	High-performance liquid chromatography: purification and chromatographic behaviour of molecular variants of pepsinogen A from human urine. *Biomedical Applications*, **1991**, 571, 47-59		3
13	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, 101-5	3.6	1
12	Human pepsinogen A isozymogen patterns in serum and gastric mucosa. *Gastroenterology*, **1990**, 99, 1576-80	13.3	7
11	Human pepsinogen C (progastricsin) polymorphism: evidence for a single locus located at 6p21.1-pter. *Genomics*, **1989**, 4, 137-48	4.3	39
10	Immunohistochemical localization of pepsinogen A and C containing cells in Barrett oesophagus. *Virchows Archiv A, Pathological Anatomy and Histopathology*, **1988**, 413, 11-6		1
9	Effect of high dose omeprazole on gastric pepsin secretion and serum pepsinogen levels in man. *European Journal of Clinical Pharmacology*, **1988**, 35, 173-6	2.8	30
8	Discrepancies between gastric mucosal and urinary pepsinogen A patterns and in vitro synthesis and secretion of human pepsinogen. *Digestive Diseases and Sciences*, **1988**, 33, 135-43	4	8
7	RFLP for the human pepsinogen C gene (PGC). *Nucleic Acids Research*, **1988**, 16, 9372	20.1	12
6	Renal handling of pepsinogens A and C in man. *Clinical Science*, **1988**, 75, 649-54	6.5	21
5	Enzyme-linked immunosorbent assay and radioimmunoassay of serum pepsinogen A. *Scandinavian Journal of Clinical and Laboratory Investigation*, **1987**, 47, 29-33	2	11
4	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, 561-6	4	52
3	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, 617-23	4	6
2	The influence of omeprazole on the synthesis and secretion of pepsinogen in isolated rabbit gastric glands. *Biochemical Pharmacology*, **1985**, 34, 3693-9	6	10
1	Genetics of urinary pepsinogen: a new hypothesis. *Human Genetics*, **1984**, 65, 385-90	6.3	44