Constantinos Deltas

List of Publications by Year
in descending order

129 papers	6,895 citations	81900 39 h-index	64796 79 g-index
132 all docs	132 docs citations	132 times ranked	6588 citing authors

#	Article	IF	Citations
1	Genotypeâ€“phenotype correlations for COL4A3â€“COL4A5 variants resulting in Gly substitutions in Alport syndrome. Scientific Reports, 2022, 12, 2722.	3.3	21
2	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	2.8	12
3	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	4.5	49
4	Mild X-linked Alport syndrome due to the COL4A5 G624D variant originating in the Middle Ages is predominant in Central/East Europe and causes kidney failure in midlife. Kidney International, 2021, 99, 1451-1458.	5.2	21
5	A glycine substitution in the collagenous domain of Col4a3 in mice recapitulates late onset Alport syndrome. Matrix Biology Plus, 2021, 9, 100053.	3.5	3
6	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	2.8	61
7	Evidence for miR-548c-5p regulation of FOXC2 transcription through a distal genomic target site in human podocytes. Cellular and Molecular Life Sciences, 2020, 77, 2441-2459.	5.4	7
8	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27
9	Prevalence of clinical, pathological and molecular features of glomerular basement membrane nephropathy caused byCOL4A3orCOL4A4mutations: a systematic review. CKJ: Clinical Kidney Journal, 2020, 13, 1025-1036.	2.9	51
10	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease due to mutationsÂin UMOD and MUC1. Kidney International, 2020, 98, 717-731.	5.2	75
11	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	1.7	97
12	Small Molecule Targets TMED9 and Promotes Lysosomal Degradation to Reverse Proteinopathy. Cell, 2019, 178, 521-535.e23.	28.9	124
13	Clinical course and outcome after kidney transplantation in patients with C3 glomerulonephritis due to CFHR5 nephropathy. Nephrology Dialysis Transplantation, 2019, 34, 1780-1788.	0.7	5
14	Omega-3 fatty acids protect retinal neurons in the DBA/2J hereditary glaucoma mouse model. Experimental Eye Research, 2018, 167, 128-139.	2.6	23
15	Distal renal tubular acidosis in a Libyan patient: Evidence for digenic inheritance. European Journal of Medical Genetics, 2018, 61, 1-7.	1.3	7
16	Digenic inheritance and genetic modifiers. Clinical Genetics, 2018, 93, 429-438.	2.0	75
17	COL4A5 and LAMA5 variants co-inherited in familial hematuria: digenic inheritance or genetic modifier effect?. BMC Nephrology, 2018, 19, 114.	1.8	35
18	Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. Nephrology Dialysis Transplantation, 2017, 32, gfw095.	0.7	40

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19	Frequent <scp>COL4</scp> mutations in familial microhematuria accompanied by laterâ€onset Alport nephropathy due to focal segmental glomerulosclerosis. Clinical Genetics, 2017, 92, 517-527.	2.0	35
20	Therapeutic potential of omega-3 fatty acids supplementation in a mouse model of dry macular degeneration. BMJ Open Ophthalmology, 2017, 1, e000056.	1.6	13
21	A functional variant in NEPH3 gene confers high risk of renal failure in primary hematuric glomerulopathies. Evidence for predisposition to microalbuminuria in the general population. PLoS ONE, 2017, 12, e0174274.	2.5	20
22	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	2.5	96
23	RAAS inhibition and the course of Alport syndrome. Pharmacological Research, 2016, 107, 205-210.	7.1	18
24	Y-chromosome phylogeographic analysis of the Greek-Cypriot population reveals elements consistent with Neolithic and Bronze Age settlements. Investigative Genetics, 2016, 7, 1.	3.3	20
25	Molecular and Clinical Investigation of Cystinuria in the Greek-Cypriot Population. Genetic Testing and Molecular Biomarkers, 2015, 19, 641-645.	0.7	1
26	Autosomal dominant tubulointerstitial kidney disease: diagnosis, classification, and managementâ€”A KDIGO consensus report. Kidney International, 2015, 88, 676-683.	5.2	276
27	Co-Inheritance of Functional Podocin Variants with Heterozygous Collagen IV Mutations Predisposes to Renal Failure. Nephron, 2015, 130, 200-212.	1.8	21
28	Haploinsufficiency of the miR-873/miR-876 microRNA cluster is associated with craniofacial abnormalities. Gene, 2015, 561, 95-100.	2.2	15
29	Carriers of Autosomal Recessive Alport Syndrome with Thin Basement Membrane Nephropathy Presenting as Focal Segmental Glomerulosclerosis in Later Life. Nephron, 2015, 130, 271-280.	1.8	55
30	COL4A3/COL4A4 heterozygous mutations with TBMN presenting as focal segmental glomerulosclerosis. Kidney International, 2015, 87, 859.	5.2	5
31	New miRNA Profiles Accurately Distinguish Renal Cell Carcinomas and Upper Tract Urothelial Carcinomas from the Normal Kidney. PLoS ONE, 2014, 9, e91646.	2.5	42
32	ccRCC is fundamentally a metabolic disorder. Cell Cycle, 2014, 13, 2481-2482.	2.6	6
33	A novel splice-site mutation in ATP6V0A4 gene in two brothers with distal renal tubular acidosis from a consanguineous Tunisian family. Journal of Genetics, 2014, 93, 859-863.	0.7	3
34	Variable Clinical Presentation of an MUC1 Mutation Causing Medullary Cystic Kidney Disease Type 1. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 527-535.	4.5	65
35	DNA variant databases improve test accuracy and phenotype prediction in Alport syndrome. Pediatric Nephrology, 2014, 29, 971-977.	1.7	22
36	Alport syndrome from bench to bedside: the potential of current treatment beyond RAAS blockade and the horizon of future therapies. Nephrology Dialysis Transplantation, 2014, 29, iv124-iv130.	0.7	38

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37	Molecular Investigation of Distal Renal Tubular Acidosis in Tunisia, Evidence for Founder Mutations. Genetic Testing and Molecular Biomarkers, 2014, 18, 741-748.	0.7	15
38	Evidence for Activation of the Unfolded Protein Response in Collagen IV Nephropathies. Journal of the American Society of Nephrology: JASN, 2014, 25, 260-275.	6.1	71
39	Genetic polymorphisms in warfarin and tacrolimus-related genes VKORC1, CYP2C9 and CYP3A5 in the Greek-Cypriot population. BMC Research Notes, 2014, 7, 123.	1.4	6
40	Frequency of COL4A3/COL4A4 Mutations amongst Families Segregating Glomerular Microscopic Hematuria and Evidence for Activation of the Unfolded Protein Response. Focal and Segmental Glomerulosclerosis Is a Frequent Development during Ageing. PLoS ONE, 2014, 9, e115015.	2.5	53
41	Altered metabolic pathways in clear cell renal cell carcinoma: A meta-analysis and validation study focused on the deregulated genes and their associated networks.. Oncoscience, 2014, 1, 117-131.	2.2	42
42	A mathematical model of the unfolded protein stress response reveals the decision mechanism for recovery, adaptation and apoptosis. BMC Systems Biology, 2013, 7, 16.	3.0	46
43	Molecular genetics of familial hematuric diseases. Nephrology Dialysis Transplantation, 2013, 28, 2946-2960.	0.7	63
44	C3 Glomerulonephritis/CFHR5 Nephropathy Is an Endemic Disease in Cyprus: Clinical and Molecular Findings in 21 Families. Advances in Experimental Medicine and Biology, 2013, 735, 189-196.	1.6	25
45	On â€˜Incidence of renal failure and nephroprotection by RAAS inhibition in heterozygous carriers of X-chromosomal and autosomal recessive Alport mutationsâ€™. Kidney International, 2013, 83, 331.	5.2	2
46	Epistatic Role of the MYH9/APOL1 Region on Familial Hematuria Genes. PLoS ONE, 2013, 8, e57925.	2.5	11
47	CNVs-microRNAs Interactions Demonstrate Unique Characteristics in the Human Genome. An Interspecies in silico Analysis. PLoS ONE, 2013, 8, e81204.	2.5	15
48	Renal graft outcome in autosomal dominant medullary cystic kidney disease type 1. Journal of Nephrology, 2013, 26, 793-798.	2.0	4
49	X-linked, COL4A5 hypomorphic Alport mutations such as G624D and P628L may only exhibit thin basement membrane nephropathy with microhematuria and late onset kidney failure. Hippokratia, 2013, 17, 207-13.	0.3	30
50	Is suppression of cyst growth in PKD enough to preserve renal function?. Jak-stat, 2012, 1, 216-218.	2.2	3
51	Genetic diseases. Nephrology Dialysis Transplantation, 2012, 27, ii320-ii329.	0.7	3
52	Genotypeâ€“phenotype correlation in Xâ€linked Alport syndrome patients carrying missense mutations in the collagenous domain of COL4A5. Clinical Genetics, 2012, 82, 297-299.	2.0	20
53	A miR-1207-5p Binding Site Polymorphism Abolishes Regulation of HBEGF and Is Associated with Disease Severity in CFHR5 Nephropathy. PLoS ONE, 2012, 7, e31021.	2.5	47
54	The role of molecular genetics in diagnosing familial hematuria(s). Pediatric Nephrology, 2012, 27, 1221-1231.	1.7	39

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55	Xâ€linked Alport syndrome in Hellenic families: Phenotypic heterogeneity and mutations near interruptions of the collagen domain in COL4A5. Clinical Genetics, 2012, 81, 240-248.	2.0	38
56	Evidence that NPHS2-R229Q predisposes to proteinuria and renal failure in familial hematuria. Pediatric Nephrology, 2012, 27, 675-679.	1.7	51
57	Increased Number of MicroRNA Target Sites in Genes Encoded in CNV Regions. Evidence for an Evolutionary Genomic Interaction. Molecular Biology and Evolution, 2011, 28, 2421-2424.	8.9	29
58	Familial C3 Glomerulopathy Associated with CFHR5 Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1436-1446.	4.5	124
59	Founder Mutations in the ATP6V1B1 GeneExplain Most Cypriot Cases of Distal Renal Tubular Acidosis: First Prenatal Diagnosis. Nephron Clinical Practice, 2011, 117, c206-c212.	2.3	6
60	RET proto-oncogene mutations are restricted to codon 618 in Cypriot families with multiple endocrine neoplasia 2. Journal of Endocrinological Investigation, 2011, 34, 764-9.	3.3	4
61	Cyst formation in the PKD2 (1-703) transgenic rat precedes deregulation of proliferation-related pathways. BMC Nephrology, 2010, 11, 23.	1.8	13
62	Genetic Variation of DKK3 May Modify Renal Disease Severity in ADPKD. Journal of the American Society of Nephrology: JASN, 2010, 21, 1510-1520.	6.1	59
63	SURVEYOR on the Spot. Journal of Molecular Diagnostics, 2010, 12, 265-266.	2.8	1
64	Identification of a mutation in complement factor H-related protein 5 in patients of Cypriot origin with glomerulonephritis. Lancet, The, 2010, 376, 794-801.	13.7	298
65	Cystic Diseases of the Kidney: Molecular Biology and Genetics. Archives of Pathology and Laboratory Medicine, 2010, 134, 569-582.	2.5	31
66	microRNAs: a newly described class of encoded molecules that play a role in health and disease. Hippokratia, 2010, 14, 236-40.	0.3	279
67	Screening for Familial Mediterranean Fever M694V and V726A Mutations in the Greek Population. Genetic Testing and Molecular Biomarkers, 2009, 13, 291-293.	0.7	4
68	Clinico-pathological correlations in 127 patients in 11 large pedigrees, segregating one of three heterozygous mutations in the COL4A3/ COL4A4 genes associated with familial haematuria and significant late progression to proteinuria and chronic kidney disease from focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2009, 24, 2721-2729.	0.7	129
69	Thin basement membrane nephropathy: is there genetic predisposition to more severe disease?. Pediatric Nephrology, 2009, 24, 877-879.	1.7	8
70	2nd Combined Working Group and Management Committee Meeting of Urine and Kidney Proteomics COST Action 29â€“30 March 2009, Nafplio, Greece. Proteomics - Clinical Applications, 2009, 3, 1017-1022.	1.6	9
71	Screening for Mutations in Kidney-Related Genes Using SURVEYOR Nuclease for Cleavage at Heteroduplex Mismatches. Journal of Molecular Diagnostics, 2009, 11, 311-318.	2.8	21
72	Serum total homocysteine, folate, 5,10-methylenetetrahydrofolate reductase (MTHFR) 677Câ†'T genotype and subclinical atherosclerosis. Expert Opinion on Therapeutic Targets, 2009, 13, 1-11.	3.4	13

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73	NPHS2 screening with SURVEYOR in Hellenic children with steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2008, 23, 1373-1375.	1.7	10
74	Mutant polycystin-2 induces proliferation in primary rat tubular epithelial cells in a STAT-1/p21-independent fashion accompanied instead by alterations in expression of p57KIP2and Cdk2. BMC Nephrology, 2008, 9, 10.	1.8	22
75	COL4A3Founder Mutations in Greek-Cypriot Families with Thin Basement Membrane Nephropathy and Focal Segmental Glomerulosclerosis Dating from Around 18th Century. Genetic Testing and Molecular Biomarkers, 2008, 12, 273-278.	1.7	25
76	COL4A3/COL4A4Mutations Link Familial Hematuria and Focal Segmental Glomerulosclerosis. Glomerular Epithelium Destruction via Basement Membrane Thinning?. Connective Tissue Research, 2008, 49, 283-288.	2.3	30
77	Lack of association between endothelial nitric oxide synthase gene polymorphisms and risk of premature coronary artery disease in the Greek population. Acta Cardiologica, 2008, 63, 609-614.	0.9	23
78	COL4A3/COL4A4 Mutations Producing Focal Segmental Glomerulosclerosis and Renal Failure in Thin Basement Membrane Nephropathy. Journal of the American Society of Nephrology: JASN, 2007, 18, 3004-3016.	6.1	187
79	Analysis of published PKD1 gene sequence variants. Nature Genetics, 2007, 39, 427-428.	21.4	19
80	Cystic fibrosis mutational spectrum and genotypic/phenotypic features in Greek-Cypriots, with emphasis on dehydration as presenting symptom. Clinical Genetics, 2007, 71, 290-292.	2.0	19
81	Molecular investigation and longâ€term clinical progress in Greek Cypriot families with recessive distal renal tubular acidosis and sensorineural deafness due to mutations in the ATP6V1B1 gene. Clinical Genetics, 2006, 69, 135-144.	2.0	24
82	Medullary cystic kidney disease type 1: mutational analysis in 37 genes based on haplotype sharing. Human Genetics, 2006, 119, 649-658.	3.8	34
83	The Cypriot and Iranian National Mutation Frequency Databases. Human Mutation, 2006, 27, 598-599.	2.5	32
84	RNA Intereference: a powerful laboratory tool and its therapeutic implications. Hippokratia, 2006, 10, 112-5.	0.3	6
85	Multiplex Molecular Diagnosis of MEFV Mutations in Patients with Familial Mediterranean Fever by LightCycler Real-Time PCR. Clinical Chemistry, 2005, 51, 1725-1727.	3.2	5
86	The MTHFR 677TT and 677CT/1298AC genotypes in Cypriot patients may be predisposing to hypertensive nephrosclerosis and chronic renal failure. International Angiology, 2005, 24, 287-94.	0.9	19
87	Modification of the enzyme mismatch cleavage method using T7 endonuclease I and silver staining. BioTechniques, 2004, 36, 758-760.	1.8	11
88	Description of the First Two Seemingly Unrelated Greek Cypriot Families with a Common C618R RET Proto-Oncogene Mutation. Genetic Testing and Molecular Biomarkers, 2004, 8, 163-168.	1.7	8
89	Non-isotopic RNase cleavage assay for mutation detection in MEFV, the gene responsible for familial Mediterranean fever, in a cohort of Greek patients. Annals of the Rheumatic Diseases, 2004, 63, 438-443.	0.9	37
90	Evidence for Association of Endothelial Cell Nitric Oxide Synthase Gene Polymorphism with Earlier Progression to End-Stage Renal Disease in a Cohort of Hellens from Greece and Cyprus. Genetic Testing and Molecular Biomarkers, 2004, 8, 319-324.	1.7	24

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91	Discovery of old diseases: the molecular approach. European Journal of Human Genetics, 2003, 11, 3-4.	2.8	3
92	Familial Mediterranean fever associated pyrin mutations in Greece. Annals of the Rheumatic Diseases, 2003, 62, 479-481.	0.9	52
93	Genotype-Renal Function Correlation in Type 2 Autosomal Dominant Polycystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2003, 14, 1164-1174.	6.1	129
94	Outcome of kidney transplantation in autosomal dominant medullary cystic kidney disease type 1. Nephrology Dialysis Transplantation, 2003, 18, 2165-2169.	0.7	18
95	Familial Mediterranean Fever (FMF) Mutations Occur Frequently in the Greek-Cypriot Population of Cyprus. Genetic Testing and Molecular Biomarkers, 2002, 6, 15-21.	1.7	40
96	A family with the branchio-oto-renal syndrome: clinical and genetic correlations. Nephrology Dialysis Transplantation, 2002, 17, 1014-1018.	0.7	30
97	Autosomal-dominant medullary cystic kidney disease type 1: Clinical and molecular findings in six large Cypriot families. Kidney International, 2002, 62, 1385-1394.	5.2	65
98	Refinement of the Gene Locus for Autosomal Dominant Medullary Cystic Kidney Disease Type 1 (MCKD1) and Construction of a Physical and Partial Transcriptional Map of the Region. Genomics, 2001, 72, 278-284.	2.9	29
99	Novel NPR1 polymorphic variants and its exclusion as a candidate gene for medullary cystic kidney disease (ADMCKD) type 1. Molecular and Cellular Probes, 2001, 15, 357-361.	2.1	7
100	Mutations of the human polycystic kidney disease 2 (PKD2) gene. Human Mutation, 2001, 18, 13-24.	2.5	49
101	Novel PKD1 deletions and missense variants in a cohort of Hellenic polycystic kidney disease families. European Journal of Human Genetics, 2001, 9, 677-684.	2.8	13
102	Autosomal dominant polycystic kidney disease: molecular genetics and molecular pathogenesis. Human Genetics, 2000, 107, 115-126.	3.8	50
103	Autosomal dominant polycystic kidney diseaseâ€"type 2. Ultrasound, genetic and clinical correlations. Nephrology Dialysis Transplantation, 2000, 15, 205-211.	0.7	51
104	Genetic evidence for a trans-heterozygous model for cystogenesis in autosomal dominant polycystic kidney disease. Human Molecular Genetics, 2000, 9, 447-452.	2.9	92
105	Prevalence of Genetic Mutations That Predispose to Thrombophilia in a Greek Cypriot Population. Clinical and Applied Thrombosis/Hemostasis, 2000, 6, 104-107.	1.7	32
106	Germinal and somatic mutations in the PKD2 gene of renal cysts in autosomal dominant polycystic kidney disease. Human Molecular Genetics, 1999, 8, 509-513.	2.9	88
107	Loss of heterozygosity in polycystic kidney disease with a missense mutation in the repeated region of PKD1. Human Genetics, 1998, 103, 709-717.	3.8	57
108	Chromosome 1 localization of a gene for autosomal dominant medullary cystic kidney disease. Human Molecular Genetics, 1998, 7, 905-911.	2.9	122

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109	Autosomal dominant medullary cystic kidney disease: evidence of gene locus heterogeneity. Nephrology Dialysis Transplantation, 1998, 13, 1955-1957.	0.7	28
110	A Translation Frameshift Mutation Induced by a Cytosine Insertion in the Polycystic Kidney Disease 2 Gene (PKD2). Human Molecular Genetics, 1997, 6, 949-952.	2.9	30
111	Clinical Aspects of Cystinuria. , 1997, 122, 167-172.		7
112	New amino acid polymorphism, Ala/Val4058, in exon 45 of the polycystic kidney disease 1 gene: evolution of alleles. Human Genetics, 1997, 99, 644-647.	3.8	11
113	Description of a symptomless cystic fibrosis L346P/M348K compound heterozygous Cypriot individual. Molecular and Cellular Probes, 1996, 10, 315-318.	2.1	9
114	PKD2 , a Gene for Polycystic Kidney Disease That Encodes an Integral Membrane Protein. Science, 1996, 272, 1339-1342.	12.6	1,303
115	Complete coding sequence, exon/intron arrangement and chromosome location of ZNF45, a KRAB-domain-containing gene. Cytogenetic and Genome Research, 1996, 75, 230-233.	1.1	8
116	Detection of a novel nonsense mutation and an intragenic polymorphism in the PKD1 gene of a Cypriot family with autosomal dominant polycystic kidney disease. Human Genetics, 1996, 98, 437-442.	3.8	37
117	Genetic heterogeneity in adult dominant polycystic kidney disease in Cypriot families. Human Genetics, 1995, 95, 416-23.	3.8	14
118	A base substitution at IVS-19 3?-end splice junction causes exon 20 skipping in pro?2(I) collagen mRNA and produces mild osteogenesis imperfecta. Human Genetics, 1994, 93, 681-7.	3.8	14
119	Novel cystic fibrosis mutation associated with mild disease in Cypriot patients. Human Genetics, 1994, 93, 529-32.	3.8	3
120	Cystic fibrosis patients from the black sea region: The 1677delTA mutation. Human Mutation, 1994, 3, 353-357.	2.5	16
121	Somatic cell mosaicism: Another source of phenotypic heterogeneity in nuclear families with osteogenesis imperfecta. American Journal of Medical Genetics Part A, 1993, 45, 246-251.	2.4	23
122	Chromosome 4 localization of a second gene for autosomal dominant polycystic kidney disease. Nature Genetics, 1993, 5, 359-362.	21.4	272
123	The identification and characterization of KRAB-domain-containing zinc finger proteins. Genomics, 1992, 12, 581-589.	2.9	21
124	An error in dystrophin mRNA processing in golden retriever muscular dystrophy, an animal homologue of Duchenne muscular dystrophy. Genomics, 1992, 13, 115-121.	2.9	312
125	G to A polymorphism in exon 45 of the COL1A1 gene. Nucleic Acids Research, 1991, 19, 4302-4302.	14.5	6
126	Mutation in a gene for type I procollagen (COL1A2) in a woman with postmenopausal osteoporosis: evidence for phenotypic and genotypic overlap with mild osteogenesis imperfecta.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 5423-5427.	7.1	138

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127	A Substitution of Cysteine for Glycine 904 in COL1A1 in a Proband with Lethal Osteogenesis Imperfecta and in Her Asymptomatic Mother. Annals of the New York Academy of Sciences, 1990, 580, 540-541.	3.8	0
128	A lethal variant of osteogenesis imperfecta has a single base mutation that substitutes cysteine for glycine 904 of the alpha 1(I) chain of type I procollagen. The asymptomatic mother has an unidentified mutation producing an overmodified and unstable type I procollagen.. Journal of Clinical Investigation, 1989, 83, 574-584.	8.2	59
129	The A and B fragments of normal type I procollagen have a similar thermal stability to proteinase digestion but are selectively destabilized by structural mutations. FEBS Journal, 1987, 163, 247-251.	0.2	20