

Chapter 137

Lipoid Proteinosis and Heritable Disorders of Connective Tissue

Jonathan A. Dyer

REFERENCES

1. Beighton P et al: Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK). *Am J Med Genet* **77**(1):31-37, 1998
2. Furukawa K, Okajima T: Galactosyltransferase I is a gene responsible for progeroid variant of Ehlers-Danlos syndrome: Molecular cloning and identification of mutations. *Biochim Biophys Acta* **1573**(3):377-381, 2002
3. Beighton P: Hypermobility scoring. *Br J Rheumatol* **27**(2):163, 1988
4. Lawrence EJ: The clinical presentation of Ehlers-Danlos syndrome. *Adv Neonatal Care* **5**(6):301-314, 2005
5. Wenstrup RJ et al: COL5A1 haploinsufficiency is a common molecular mechanism underlying the classical form of EDS. *Am J Hum Genet* **66**(6):1766-1776, 2000
6. Symoens S et al: COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. *Hum Mutat* **30**(2):E395-E403, 2009
7. Wenstrup RJ et al: A splice-junction mutation in the region of COL5A1 that codes for the carboxyl propeptide of pro alpha 1(V) chains results in the gravis form of the Ehlers-Danlos syndrome (type I). *Hum Mol Genet* **5**(11):1733-1736, 1996
8. Greenspan DS et al: COL5A1: Fine genetic mapping and exclusion as candidate gene in families with nail-patella syndrome, tuberous sclerosis 1, hereditary hemorrhagic telangiectasia, and Ehlers-Danlos Syndrome type II. *Genomics* **25**(3):737-739, 1995
9. Nuytinck L et al: Classical Ehlers-Danlos syndrome caused by a mutation in type I collagen. *Am J Hum Genet* **66**(4):1398-1402, 2000
10. Malfait F et al: Three arginine to cysteine substitutions in the pro-alpha (I)-collagen chain cause Ehlers-Danlos syndrome with a propensity to arterial rupture in early adulthood. *Hum Mutat* **28**(4):387-395, 2007
11. Schwarze U et al: Rare autosomal recessive cardiac valvular form of Ehlers-Danlos syndrome results from mutations in the COL1A2 gene that activate the nonsense-mediated RNA decay pathway. *Am J Hum Genet* **74**(5):917-930, 2004
12. Burch GH et al: Tenascin-X deficiency is associated with Ehlers-Danlos syndrome. *Nat Genet* **17**(1):104-108, 1997
13. Mao JR, Bristow J: The Ehlers-Danlos syndrome: on beyond collagens. *J Clin Invest* **107**(9):1063-1069, 2001
14. Hausser I, Anton-Lamprecht I: Differential ultrastructural aberrations of collagen fibrils in Ehlers-Danlos syndrome types I-IV as a means of diagnostics and classification. *Hum Genet* **93**(4):394-407, 1994
15. Grahame R, Beighton P: Physical properties of the skin in the Ehlers-Danlos syndrome. *Ann Rheum Dis* **28**(3):246-251, 1969
16. Tiller GE et al: Aortic root dilatation in Ehlers-Danlos syndrome types I, II and III. A report of five cases. *Clin Genet* **53**(6):460-465, 1998
17. Machet L et al: Absence of inferior labial and lingual frenula in Ehlers-Danlos syndrome: A minor diagnostic criterion in French patients. *Am J Clin Dermatol* **11**(4):269-273, 2010
18. Pepin M et al: Clinical and genetic features of Ehlers-Danlos syndrome type IV, the vascular type. *N Engl J Med* **342**(10):673-680, 2000
19. Wenstrup R, De PA: Ehlers-Danlos syndrome, classic type. In: GeneReviews at GeneTests: Medical Genetics Information Resource. Seattle, University of Washington, 1993, <http://www.genetests.org>
20. Snyder RR, Gilstrap LC, Hauth JC: Ehlers-Danlos syndrome and pregnancy. *Obstet Gynecol* **61**(5):649-650, 1983
21. Ramos-E-Silva M et al: Connective tissue diseases: Pseudoxanthoma elasticum, anetoderma, and Ehlers-Danlos syndrome in pregnancy. *Clin Dermatol* **24**(2):91-96, 2006
22. Wenstrup RJ et al: Prevalence of aortic root dilatation in the Ehlers-Danlos syndrome. *Genet Med* **4**(3):112-117, 2002

23. Narcisi P et al: A family with Ehlers-Danlos syndrome type III/articular hypermobility syndrome has a glycine 637 to serine substitution in type III collagen. *Hum Mol Genet* 3(9):1617-1620, 1994
24. Zweers MC et al: Haploinsufficiency of TNXB is associated with hypermobility type of Ehlers-Danlos syndrome. *Am J Hum Genet* 73(1):214-217, 2003
25. Callewaert B et al: Ehlers-Danlos syndromes and Marfan syndrome. *Best Pract Res Clin Rheumatol* 22(1):165-189, 2008
26. Chen W et al: The phenotypic spectrum of contiguous deletion of CYP21A2 and tenascin XB: Quadricuspid aortic valve and other midline defects. *Am J Med Genet A* 149A(12):2803-2808, 2009
27. Grahame R: Heritable disorders of connective tissue. *Baillieres Best Pract Res Clin Rheumatol* 14(2):345-361, 2000
28. Harinsein D et al: Systemic joint laxity (the hypermobile joint syndrome) is associated with temporomandibular joint dysfunction. *Arthritis Rheum* 31(10):1259-1264, 1988
29. Stoler JM, Oaklander AL: Patients with Ehlers Danlos syndrome and CRPS: A possible association? *Pain* 123(1-2):204-209, 2006
30. Lumley MA et al: Psychosocial functioning in the Ehlers-Danlos syndrome. *Am J Med Genet* 53(2):149-152, 1994
31. Levy HP: Ehlers-Danlos syndrome, hypermobility type. In: GeneReviews at GeneTests: Medical Genetics Information Resource. Seattle, University of Washington, 1993, <http://www.genetests.org>
32. Kobayasi T: Dermal elastic fibres in the inherited hypermobile disorders. *J Dermatol Sci* 41(3):175-185, 2006
33. McDonnell NB et al: Echocardiographic findings in classical and hypermobile Ehlers-Danlos syndromes. *Am J Med Genet A* 140(2):129-136, 2006
34. Castori M et al: Natural history and manifestations of the hypermobility type Ehlers-Danlos syndrome: A pilot study on 21 patients. *Am J Med Genet A* 152A(3):556-564, 2010
35. Cole WG et al: A base substitution at a splice site in the COL3A1 gene causes exon skipping and generates abnormal type III procollagen in a patient with Ehlers-Danlos syndrome type IV. *J Biol Chem* 265(28):17070-17077, 1990
36. Germain DP: Clinical and genetic features of vascular Ehlers-Danlos syndrome. *Ann Vasc Surg* 16(3):391-397, 2002
37. Pinnell SR et al: A heritable disorder of connective tissue. Hydroxylysine-deficient collagen disease. *N Engl J Med* 286(19):1013-1020, 1972
38. Eyre DR, Glimcher MJ: Reducible crosslinks in hydroxylysine-deficient collagens of a heritable disorder of connective tissue. *Proc Natl Acad Sci U S A* 69(9):2594-2598, 1972
39. Heikkinen J et al: Duplication of seven exons in the lysyl hydroxylase gene is associated with longer forms of a repetitive sequence within the gene and is a common cause for the type VI variant of Ehlers-Danlos syndrome. *Am J Hum Genet* 60(1):48-56, 1997
40. Ogur G et al: Clinical, ultrastructural and biochemical studies in two sibs with Ehlers-Danlos syndrome type VI-B-like features. *Clin Genet* 46(6):417-422, 1994
41. Pasquali M et al: Urinary pyridinium cross-links: A noninvasive diagnostic test for Ehlers-Danlos syndrome type VI. *N Engl J Med* 331(2):132-133, 1994
42. Eyre D et al: The kyphoscoliotic type of Ehlers-Danlos syndrome (type VI): Differential effects on the hydroxylation of lysine in collagens I and II revealed by analysis of cross-linked telopeptides from urine. *Mol Genet Metab* 76(3):211-216, 2002
43. Giunta C et al: The arthrochalasia type of Ehlers-Danlos syndrome (EDS VIIA and VIIB): The diagnostic value of collagen fibril ultrastructure. *Am J Med Genet A* 146A(10):1341-1346, 2008
44. Byers PH et al: Ehlers-Danlos syndrome type VIIA and VIIB result from splice-junction mutations or genomic deletions that involve exon 6 in the COL1A1 and COL1A2 genes of type I collagen. *Am J Med Genet* 72(1):94-105, 1997
45. Ho KK et al: Further evidence that the failure to cleave the aminopropeptide of type I procollagen is the cause of Ehlers-Danlos syndrome type VII. *Hum Mutat* 3(4):358-364, 1994
46. Nicholls AC et al: Clinical phenotypes and molecular characterisation of three patients with Ehlers-Danlos syndrome type VII. *J Med Genet* 37(11):E33, 2000
47. Giunta C et al: Ehlers-Danlos syndrome type VII: Clinical features and molecular defects. *J Bone Joint Surg Am* 81(2):225-238, 1999
48. Lehmann HW et al: Ehlers-Danlos syndrome type VII: Phenotype and genotype. *Arch Dermatol Res* 286(8):425-428, 1994
49. Colige A et al: Human Ehlers-Danlos syndrome type VII C and bovine dermatosparaxis are caused by mutations in the procollagen I N-proteinase gene. *Am J Hum Genet* 65(2):308-317, 1999

50. Colige A et al: Novel types of mutation responsible for the dermatosparactic type of Ehlers-Danlos syndrome (Type VIIC) and common polymorphisms in the ADAMTS2 gene. *J Invest Dermatol* **123**(4):656-663, 2004
51. Smith LT et al: Human dermatosparaxis: A form of Ehlers-Danlos syndrome that results from failure to remove the amino-terminal propeptide of type I procollagen. *Am J Hum Genet* **51**(2):235-244, 1992
52. Petty EM et al: Dermatosparaxis in children. A case report and review of the newly recognized phenotype. *Arch Dermatol* **129**(10):1310-1315, 1993
53. Fujimoto A, Wilcox WR, Cohn DH: Clinical, morphological, and biochemical phenotype of a new case of Ehlers-Danlos syndrome type VIIC. *Am J Med Genet* **68**(1):25-28, 1997
54. Coman DJ: Diagnostic dilemma's: the congenital disorders of glycosylation are clinical chameleons. *Eur J Hum Genet* **16**(1):2-4, 2008
55. Makareeva E et al: Molecular mechanism of alpha 1(I)-osteogenesis imperfecta/Ehlers-Danlos syndrome: Unfolding of an N-anchor domain at the N-terminal end of the type I collagen triple helix. *J Biol Chem* **28**(10):6463-6470, 2006
56. Okajima T et al: Molecular basis for the progeroid variant of Ehlers-Danlos syndrome. Identification and characterization of two mutations in galactosyltransferase I gene. *J Biol Chem* **274**(41):28841-28844, 1999
57. Sheen VL et al: Filamin A mutations cause periventricular heterotopia with Ehlers-Danlos syndrome. *Neurology* **64**(2):254-262, 2005
58. Kosho T et al: A new Ehlers-Danlos syndrome with craniofacial characteristics, multiple congenital contractures, progressive joint and skin laxity, and multisystem fragility-related manifestations. *Am J Med Genet A* **152A**(6):1333-1346, 2010
59. Giunta C et al: Spondylocheiro dysplastic form of the Ehlers-Danlos syndrome—an autosomal-recessive entity caused by mutations in the zinc transporter gene SLC39A13. *Am J Hum Genet* **82**(6):1290-1305, 2008
60. Loeys BL, Dietz HC: Loeys-Dietz syndrome. In: GeneReviews at GeneTests: Medical Genetics Information Resource. Seattle, University of Washington, 1993, <http://www.genetests.org>
61. Hoornaert KP et al: Stickler syndrome caused by COL2A1 mutations: Genotype-phenotype correlation in a series of 100 patients. *Eur J Hum Genet* **18**(8):872-880, 2010
62. Rose PS et al: Stickler syndrome: Clinical characteristics and diagnostic criteria. *Am J Med Genet A* **138A**(3):199-207, 2005
63. Morris CA, Mervis CB: Williams syndrome and related disorders. *Annu Rev Genomics Hum Genet* **1**:461-484, 2000
64. Pasteris NG et al: Isolation and characterization of the faciogenital dysplasia (Aarskog-Scott syndrome) gene: A putative Rho/Rac guanine nucleotide exchange factor. *Cell* **79**(4):669-678, 1994
65. Orrico A et al: Aarskog-Scott syndrome: Clinical update and report of nine novel mutations of the FGD1 gene. *Am J Med Genet A* **152A**(2):313-318, 2010
66. Syx D et al: The RIN2 syndrome: A new autosomal recessive connective tissue disorder caused by deficiency of Ras and Rab interactor 2 (RIN2). *Hum Genet* **128**(1):79-88, 2010
67. Basel-Vanagaite L et al: RIN2 deficiency results in macrocephaly, alopecia, cutis laxa, and scoliosis: MACS syndrome. *Am J Hum Genet* **85**(2):254-263, 2009
68. Whitelaw SE: Ehlers-Danlos Syndrome, classical type: Case management. *Pediatr Nurs* **29**(6):423-426, 2003
69. Whitelaw SE: Ehlers-Danlos syndrome, classical type: Case management. *Dermatol Nurs* **16**(5):433-436, 449, 2004
70. Ehlers-Danlos National Foundation. 2010, www.ednf.org
71. Brooke BS et al: Contemporary management of vascular complications associated with Ehlers-Danlos syndrome. *J Vasc Surg* **51**(1):131-138, 2010
72. Voermans NC et al: Fatigue is a frequent and clinically relevant problem in Ehlers-Danlos syndrome. *Semin Arthritis Rheum* **40**(3):267-274, 2010
73. Voermans NC et al: Neuromuscular involvement in various types of Ehlers-Danlos syndrome. *Ann Neurol* **65**(6):687-697, 2009
74. Pyeritz RE: The Marfan syndrome. *Annu Rev Med* **51**:481-510, 2000
75. Rantamaki T et al: Recurrence of Marfan syndrome as a result of parental germ-line mosaicism for an FBN1 mutation. *Am J Hum Genet* **64**(4):993-1001, 1999
76. Faivre L et al: Clinical and molecular study of 320 children with Marfan syndrome and related type I fibrillinopathies in a series of 1009 probands with pathogenic FBN1 mutations. *Pediatrics* **123**(1):391-398, 2009

77. Pearson GD et al: Report of the National Heart, Lung, and Blood Institute and National Marfan Foundation Working Group on research in Marfan syndrome and related disorders. *Circulation* **118**(7):785-791, 2008
78. Lindeman JH et al: Distinct defects in collagen microarchitecture underlie vessel-wall failure in advanced abdominal aneurysms and aneurysms in Marfan syndrome. *Proc Natl Acad Sci U S A* **107**(2):862-865, 2010
79. Dietz HC, Pyeritz RE: Mutations in the human gene for fibrillin-1 (FBN1) in the Marfan syndrome and related disorders. *Hum Mol Genet* **4**(Spec No):1799-1809, 1995
80. Dietz HC et al: Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. *Nature* **352**(6333):337-339, 1991
81. Robinson PN et al: The molecular genetics of Marfan syndrome and related disorders. *J Med Genet* **43**(10):769-787, 2006
82. Ramirez F: Fibrillin mutations in Marfan syndrome and related phenotypes. *Curr Opin Genet Dev* **6**(3):309-315, 1996
83. Goumans MJ, Liu Z, ten DP: TGF-beta signaling in vascular biology and dysfunction. *Cell Res* **19**(1):116-127, 2009
84. Jones JA, Ikonomidis JS: The pathogenesis of aortopathy in Marfan syndrome and related diseases. *Curr Cardiol Rep* **12**(2):99-107, 2010
85. Rybczynski M et al: The spectrum of syndromes and manifestations in individuals screened for suspected Marfan syndrome. *Am J Med Genet A* **146A**(24):3157-3166, 2008
86. Joseph KN et al: Orthopedic aspects of the Marfan phenotype. *Clin Orthop Relat Res* (277):251-261, 1992
87. Maumenee IH: The eye in the Marfan syndrome. *Trans Am Ophthalmol Soc* **79**:684-733, 1981
88. Jeremy RW et al: Relation between age, arterial distensibility, and aortic dilatation in the Marfan syndrome. *Am J Cardiol* **74**(4):369-373, 1994
89. Groenink M et al: Survival and complication free survival in Marfan's syndrome: Implications of current guidelines. *Heart* **82**(4):499-504, 1999
90. Silverman DI et al: Life expectancy in the Marfan syndrome. *Am J Cardiol* **75**(2):157-160, 1995
91. Seliem MA et al: Echocardiographic evaluation of the aortic root and mitral valve in children and adolescents with isolated pectus excavatum: Comparison with Marfan patients. *Pediatr Cardiol* **13**(1):20-23, 1992
92. Ho NC, Tran JR, Bektas A: Marfan's syndrome. *Lancet* **366**(9501):1978-1981, 2005
93. De PA et al: Revised diagnostic criteria for the Marfan syndrome. *Am J Med Genet* **62**(4):417-426, 1996
94. Morse RP et al: Diagnosis and management of infantile marfan syndrome. *Pediatrics* **86**(6):888-895, 1990
95. Buntinx IM et al: Neonatal Marfan syndrome with congenital arachnodactyly, flexion contractures, and severe cardiac valve insufficiency. *J Med Genet* **28**(4):267-273, 1991
96. Gupta PA et al: Ten novel FBN2 mutations in congenital contractural arachnodactyly: Delineation of the molecular pathogenesis and clinical phenotype. *Hum Mutat* **19**(1):39-48, 2002
97. Callewaert BL et al: Comprehensive clinical and molecular assessment of 32 probands with congenital contractural arachnodactyly: Report of 14 novel mutations and review of the literature. *Hum Mutat* **30**(3):334-341, 2009
98. Dietz HC. Marfan syndrome, www.ednf.org, 1993
99. Arn PH et al: Outcome of pectus excavatum in patients with Marfan syndrome and in the general population. *J Pediatr* **115**(6):954-958, 1989
100. Shores J et al: Progression of aortic dilatation and the benefit of long-term beta-adrenergic blockade in Marfan's syndrome. *N Engl J Med* **330**(19):1335-1341, 1994
101. Gray JR et al: Life expectancy in British Marfan syndrome populations. *Clin Genet* **54**(2):124-128, 1998
102. Brooke BS et al: Angiotensin II blockade and aortic-root dilation in Marfan's syndrome. *N Engl J Med* **358**(26):2787-2795, 2008
103. Hall JR et al: Pneumothorax in the Marfan syndrome: Prevalence and therapy. *Ann Thorac Surg* **37**(6):500-504, 1984
104. Yap S: Classical homocystinuria: Vascular risk and its prevention. *J Inher Metab Dis* **26**(2-3):259-265, 2003
105. Naughten ER, Yap S, Mayne PD: Newborn screening for homocystinuria: Irish and world experience. *Eur J Pediatr* **157**(Suppl. 2):S84-S87, 1998
106. Skovby F, Gaustadnes M, Mudd SH: A revisit to the natural history of homocystinuria due to cystathionine beta-synthase deficiency. *Mol Genet Metab* **99**(1):1-3, 2010

107. Munke M et al: The gene for cystathionine beta-synthase (CBS) maps to the subtelomeric region on human chromosome 21q and to proximal mouse chromosome 17. *Am J Hum Genet* **42**(4):550-559, 1988
108. Miles EW, Kraus JP: Cystathionine beta-synthase: Structure, function, regulation, and location of homocystinuria-causing mutations. *J Biol Chem* **279**(29):29871-29874, 2004
109. Kraus JP et al: Cystathionine beta-synthase mutations in homocystinuria. *Hum Mutat* **13**(5):362-375, 1999
110. Urreiziti R et al: The p.T191M mutation of the CBS gene is highly prevalent among homocystinuric patients from Spain, Portugal and South America. *J Hum Genet* **51**(4):305-313, 2006
111. Hu FL et al: Molecular basis of cystathionine beta-synthase deficiency in pyridoxine responsive and nonresponsive homocystinuria. *Hum Mol Genet* **2**(11):1857-1860, 1993
112. Gallagher PM et al: High frequency (71%) of cystathionine beta-synthase mutation G307S in Irish homocystinuria patients. *Hum Mutat* **6**(2):177-180, 1995
113. Hubmacher D et al: Functional consequences of homocysteinylation of the elastic fiber proteins fibrillin-1 and tropoelastin. *J Biol Chem* **285**(2):1188-1198, 2010
114. de Franchis R et al: Clinical aspects of cystathionine beta-synthase deficiency: How wide is the spectrum? The Italian Collaborative Study Group on Homocystinuria. *Eur J Pediatr* **157**(Suppl. 2):S67-S70, 1998
115. Mudd SH et al: The natural history of homocystinuria due to cystathionine beta-synthase deficiency. *Am J Hum Genet* **37**(1):1-31, 1985
116. Tan WH et al: Isolated sulfite oxidase deficiency: A case report with a novel mutation and review of the literature. *Pediatrics* **116**(3):757-766, 2005
117. Lerner-Ellis JP et al: Identification of the gene responsible for methylmalonic aciduria and homocystinuria, cblC type. *Nat Genet* **38**(1):93-100, 2006
118. Morel CF, Lerner-Ellis JP, Rosenblatt DS: Combined methylmalonic aciduria and homocystinuria (cblC): Phenotype-genotype correlations and ethnic-specific observations. *Mol Genet Metab* **88**(4):315-321, 2006
119. Picker JD, Levy HL: Homocystinuria caused by cystathionine beta-synthase deficiency. In: GeneReviews at GeneTests: Medical Genetics Information Resource. Seattle, University of Washington, 1993, <http://www.genetests.org>
120. Chen X et al: Contrasting behaviors of mutant cystathionine beta-synthase enzymes associated with pyridoxine response. *Hum Mutat* **27**(5):474-482, 2006
121. Lawson-Yuen A, Levy HL: The use of betaine in the treatment of elevated homocysteine. *Mol Genet Metab* **88**(3):201-207, 2006
122. Yaghmai R et al: Progressive cerebral edema associated with high methionine levels and betaine therapy in a patient with cystathionine beta-synthase (CBS) deficiency. *Am J Med Genet* **108**(1):57-63, 2002
123. Martin L et al: Heterozygosity for a single mutation in the ABCC6 gene may closely mimic PXE: Consequences of this phenotype overlap for the definition of PXE. *Arch Dermatol* **144**(3):301-306, 2008
124. Uitto J, Jiang Q: Pseudoxanthoma elasticum-like phenotypes: More diseases than one. *J Invest Dermatol* **127**(3):507-510, 2007
125. Jiang Q et al: Pseudoxanthoma elasticum is a metabolic disease. *J Invest Dermatol* **129**(2):348-354, 2009
126. Li Q et al: Pseudoxanthoma elasticum: Clinical phenotypes, molecular genetics and putative pathomechanisms. *Exp Dermatol* **18**(1):1-11, 2009
127. Le SO et al: Evidence for a founder effect for pseudoxanthoma elasticum in the Afrikaner population of South Africa. *Hum Genet* **111**(4-5):331-338, 2002
128. Jiang Q et al: Parabiogenic heterozygous pairing of Abcc6^{-/-}/Rag1^{-/-} mice and their wild-type counterparts halts ectopic mineralization in a murine model of pseudoxanthoma elasticum. *Am J Pathol* **176**(4):1855-1862, 2010
129. Vanakker OM et al: Low serum vitamin K in PXE results in defective carboxylation of mineralization inhibitors similar to the GGCC mutations in the PXE-like syndrome. *Lab Invest* **90**(6):895-905, 2010
130. Lebwohl M et al: Classification of pseudoxanthoma elasticum: Report of a consensus conference. *J Am Acad Dermatol* **30**(1):103-107, 1994
131. Kazakis AM, Parish WR: Periumbilical perforating pseudoxanthoma elasticum. *J Am Acad Dermatol* **19**(2 Pt 2):384-388, 1988
132. Lebwohl M et al: Diagnosis of pseudoxanthoma elasticum by scar biopsy in patients without characteristic skin lesions. *N Engl J Med* **317**(6):347-350, 1987

133. Hellems J et al: Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. *Nat Genet* **36**(11):1213-1218, 2004
134. Neldner KH, Martinez-Hernandez A: Localized acquired cutaneous pseudoxanthoma elasticum. *J Am Acad Dermatol* **1**(6):523-530, 1979
135. Karp DL et al: A yellow plaque with keratotic papules on the abdomen. Perforating calcific elastosis (periumbilical perforating pseudoxanthoma elasticum [PXE], localized acquired cutaneous PXE). *Arch Dermatol* **132**(2):224-228, 1996
136. Becuwe C et al: Elastosis perforans serpiginosa associated with pseudo-pseudoxanthoma elasticum during treatment of Wilson's disease with penicillamine. *Dermatology* **210**(1):60-63, 2005
137. Lewis KG et al: Acquired disorders of elastic tissue: Part II. decreased elastic tissue. *J Am Acad Dermatol* **51**(2):165-185, 2004
138. Li TH et al: An unusual cutaneous manifestation of pseudoxanthoma elasticum mimicking reticulate pigmentary disorders. *Br J Dermatol* **134**(6):1157-1159, 1996
139. Viljoen DL et al: Pseudoxanthoma elasticum in South Africa—genetic and clinical implications. *S Afr Med J* **66**(21):813-816, 1984
140. Naouri M et al: Manifestations of pseudoxanthoma elasticum in childhood. *Br J Dermatol* **161**(3):635-639, 2009
141. Georgalas I et al: Angioid streaks, clinical course, complications, and current therapeutic management. *Ther Clin Risk Manag* **5**(1):81-89, 2009
142. Nishida H et al: Coronary artery bypass in a 15-year-old girl with pseudoxanthoma elasticum. *Ann Thorac Surg* **49**(3):483-485, 1990
143. Viljoen DL, Beatty S, Beighton P: The obstetric and gynaecological implications of pseudoxanthoma elasticum. *Br J Obstet Gynaecol* **94**(9):884-888, 1987
144. Bowen AR et al: Pseudoxanthoma elasticum-like fibers in the inflamed skin of patients without pseudoxanthoma elasticum. *J Cutan Pathol* **34**(10):777-781, 2007
145. Hacker SM et al: Juvenile pseudoxanthoma elasticum: Recognition and management. *Pediatr Dermatol* **10**(1):19-25, 1993
146. Suarez MJ et al: Sonographic aspects of pseudoxanthoma elasticum. *Pediatr Radiol* **21**(7):538-539, 1991
147. Aessopos A, Farmakis D, Loukopoulos D: Elastic tissue abnormalities resembling pseudoxanthoma elasticum in beta thalassemia and the sickling syndromes. *Blood* **99**(1):30-35, 2002
148. Coatesworth AP et al: A case of systemic pseudo-pseudoxanthoma elasticum with diverse symptomatology caused by long-term penicillamine use. *J Clin Pathol* **51**(2):169-171, 1998
149. Lewis KG et al: Nephrogenic fibrosing dermopathy and calciphylaxis with pseudoxanthoma elasticum-like changes. *J Cutan Pathol* **33**(10):695-700, 2006
150. Farmakis D et al: Aortic valve replacement in a patient with thalassemia intermedia. *Ann Thorac Surg* **81**(2):737-739, 2006
151. Laube S, Moss C: Pseudoxanthoma elasticum. *Arch Dis Child* **90**(7):754-756, 2005
152. Viljoen DL, Bloch C, Beighton P: Plastic surgery in pseudoxanthoma elasticum: Experience in nine patients. *Plast Reconstr Surg* **85**(2):233-238, 1990
153. Cunningham JR et al: Pseudoxanthoma elasticum: Treatment of gastrointestinal hemorrhage by arterial embolization and observations on autosomal dominant inheritance. *Johns Hopkins Med J* **147**(4):168-173, 1980
154. Browning AC et al: Verteporfin photodynamic therapy of choroidal neovascularization in angioid streaks: One-year results of a prospective case series. *Ophthalmology* **112**(7):1227-1231, 2005
155. Lommatzsch A: Management of choroidal vascularisation. *Br J Ophthalmol* **92**(4):445-446, 2008
156. Hamamoto Y et al: Hyperreactivity of pseudoxanthoma elasticum-affected dermis to vitamin D3. *J Am Acad Dermatol* **42**(4):685-687, 2000
157. Sherer DW et al: Oral phosphate binders in the treatment of pseudoxanthoma elasticum. *J Am Acad Dermatol* **53**(4):610-615, 2005
158. Takata T et al: Treatment of pseudoxanthoma elasticum with tocopherol acetate and ascorbic acid. *Pediatr Dermatol* **24**(4):424-425, 2007
159. Li Q et al: Magnesium carbonate-containing phosphate binder prevents connective tissue mineralization in Abcc6(-/-) mice—potential for treatment of pseudoxanthoma elasticum. *Clin Transl Sci* **2**(6):398-404, 2009
160. LaRusso J et al: Elevated dietary magnesium prevents connective tissue mineralization in a mouse model of pseudoxanthoma elasticum (Abcc6(-/-)). *J Invest Dermatol* **129**(6):1388-1394, 2009

161. Graul-Neumann LM et al: Highly variable cutis laxa resulting from a dominant splicing mutation of the elastin gene. *Am J Med Genet A* **146A**(8):977-983, 2008
162. Urban Z et al: Autosomal dominant cutis laxa with severe lung disease: Synthesis and matrix deposition of mutant tropoelastin. *J Invest Dermatol* **124**(6):1193-1199, 2005
163. Beighton P: The dominant and recessive forms of cutis laxa. *J Med Genet* **9**(2):216-221, 1972
164. Damkier A, Brandrup F, Starklint H: Cutis laxa: Autosomal dominant inheritance in five generations. *Clin Genet* **39**(5):321-329, 1991
165. Markova D et al: Genetic heterogeneity of cutis laxa: A heterozygous tandem duplication within the fibulin-5 (FBLN5) gene. *Am J Hum Genet* **72**(4):998-1004, 2003
166. Kielty CM: Elastic fibres in health and disease. *Expert Rev Mol Med* **8**(19):1-23, 2006
167. Morava E et al: Autosomal recessive cutis laxa syndrome revisited. *Eur J Hum Genet* **17**(9):1099-1110, 2009
168. Ringpfeil F: Selected disorders of connective tissue: Pseudoxanthoma elasticum, cutis laxa, and lipoid proteinosis. *Clin Dermatol* **23**(1):41-46, 2005
169. Hu Q et al: Fibulin-5 mutations: Mechanisms of impaired elastic fiber formation in recessive cutis laxa. *Hum Mol Genet* **15**(23):3379-3386, 2006
170. Huchtagowder V et al: Fibulin-4: A novel gene for an autosomal recessive cutis laxa syndrome. *Am J Hum Genet* **78**(6):1075-1080, 2006
171. Reversade B et al: Mutations in PYCR1 cause cutis laxa with progeroid features. *Nat Genet* **41**(9):1016-1021, 2009
172. Morava E et al: Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. *Eur J Hum Genet* **16**(1):28-35, 2008
173. Kornak U et al: Impaired glycosylation and cutis laxa caused by mutations in the vesicular H⁺-ATPase subunit ATP6V0A2. *Nat Genet* **40**(1):32-34, 2008
174. Hennies HC et al: Geroderma osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. *Nat Genet* **40**(12):1410-1412, 2008
175. Kivuva EC et al: De Barsy syndrome: A review of the phenotype. *Clin Dysmorphol* **17**(2):99-107, 2008
176. Gupta N, Phadke SR: Cutis laxa type II and wrinkly skin syndrome: Distinct phenotypes. *Pediatr Dermatol* **23**(3):225-230, 2006
177. Urban Z et al: Mutations in LTBP4 cause a syndrome of impaired pulmonary, gastrointestinal, genitourinary, musculoskeletal, and dermal development. *Am J Hum Genet* **85**(5):593-605, 2009
178. Valayannopoulos V et al: Transaldolase deficiency: A new cause of hydrops fetalis and neonatal multi-organ disease. *J Pediatr* **149**(5):713-717, 2006
179. Bouloc A et al: Increased fibroblast elastase activity in acquired cutis laxa. *Dermatology* **198**(4):346-350, 1999
180. Hu Q et al: Inflammatory destruction of elastic fibers in acquired cutis laxa is associated with missense alleles in the elastin and fibulin-5 genes. *J Invest Dermatol* **126**(2):283-290, 2006
181. Uenishi T et al: Pseudoxanthoma elasticum with generalized cutaneous laxity. *Arch Dermatol* **133**(5):664-666, 1997
182. Vanakker OM et al: Pseudoxanthoma elasticum-like phenotype with cutis laxa and multiple coagulation factor deficiency represents a separate genetic entity. *J Invest Dermatol* **127**(3):581-587, 2007
183. Bangsgaard N et al: Nephrogenic systemic fibrosis: Late skin manifestations. *Arch Dermatol* **145**(2):183-187, 2009
184. Haider M et al: Acquired cutis laxa type II (Marshall syndrome) in an 18-month-old child: A case report. *Pediatr Dermatol* **27**(1):89-91, 2010
185. Muster AJ et al: Fatal cardiovascular disease and cutis laxa following acute febrile neutrophilic dermatosis. *J Pediatr* **102**(2):243-248, 1983
186. Hill VA, Seymour CA, Mortimer PS: Pencillamine-induced elastosis perforans serpiginosa and cutis laxa in Wilson's disease. *Br J Dermatol* **142**(3):560-561, 2000
187. Mahajan VK, Sharma NL, Garg G: Cutis laxa acquisita associated with cutaneous mastocytosis. *Int J Dermatol* **45**(8):949-951, 2006
188. Kiuru-Enari S, Keski-Oja J, Haltia M: Cutis laxa in hereditary gelsolin amyloidosis. *Br J Dermatol* **152**(2):250-257, 2005
189. Linares A et al: Reversible cutis laxa due to maternal D-penicillamine treatment. *Lancet* **2**(8132):43, 1979
190. Solomon L et al: Neonatal abnormalities associated with D-penicillamine treatment during pregnancy. *N Engl J Med* **296**(1):54-55, 1977
191. Nahas FX et al: The role of plastic surgery in congenital cutis laxa: A 10-year follow-up. *Plast Reconstr Surg* **104**(4):1174-1178, 1999

192. Fisher BK, Page E, Hanna W: Acral localized acquired cutis laxa. *J Am Acad Dermatol* **21**(1):33-40, 1989
193. Tamura BM et al: Cutis laxa: Improvement of facial aesthetics by using botulinum toxin. *Dermatol Surg* **30**(12 Pt 2):1518-1520, 2004
194. Hirano E et al: Functional rescue of elastin insufficiency in mice by the human elastin gene: Implications for mouse models of human disease. *Circ Res* **101**(5):523-531, 2007
195. Steiner RD, Pepin MG, Byers PH: Osteogenesis imperfecta. In: GeneReviews at GeneTests: Medical Genetics Information Resource. Seattle, University of Washington, 1993, <http://www.genetests.org>
196. Tinkle BT, Wenstrup RJ: A genetic approach to fracture epidemiology in childhood. *Am J Med Genet C Semin Med Genet* **139C**(1):38-54, 2005
197. Stevenson CJ, Bottoms E, Shuster S: Skin collagen in osteogenesis imperfecta. *Lancet* **1**(7652):860-861, 1970
198. Prockop DJ: Osteogenesis imperfecta: Phenotypic heterogeneity, protein suicide, short and long collagen. *Am J Hum Genet* **36**(3):499-505, 1984
199. Hansen B, Jemec GB: The mechanical properties of skin in osteogenesis imperfecta. *Arch Dermatol* **138**(7):909-911, 2002
200. Barnes AM et al: Lack of cyclophilin B in osteogenesis imperfecta with normal collagen folding. *N Engl J Med* **362**(6):521-528, 2010
201. Alanay Y et al: Mutations in the gene encoding the RER protein FKBP65 cause autosomal-recessive osteogenesis imperfecta. *Am J Hum Genet* **86**(4):551-559, 2010
202. Gahagan S, Rimsza ME: Child abuse or osteogenesis imperfecta: How can we tell? *Pediatrics* **88**(5):987-992, 1991
203. Glorieux FH: Osteogenesis imperfecta. *Best Pract Res Clin Rheumatol* **22**(1):85-100, 2008
204. Marini JC et al: The growth hormone and somatomedin axis in short children with osteogenesis imperfecta. *J Clin Endocrinol Metab* **76**(1):251-256, 1993
205. Cheung MS, Glorieux FH: Osteogenesis Imperfecta: Update on presentation and management. *Rev Endocr Metab Disord* **9**(2):153-160, 2008
206. Le BK et al: Fetal mesenchymal stem-cell engraftment in bone after in utero transplantation in a patient with severe osteogenesis imperfecta. *Transplantation* **79**(11):1607-1614, 2005
207. Gass JK et al: Buschke-Ollendorff syndrome: A manifestation of a heterozygous nonsense mutation in the LEMD3 gene. *J Am Acad Dermatol* **58**(5 Suppl. 1):S103-S104, 2008
208. Schena D et al: Buschke-Ollendorff syndrome. *Int J Dermatol* **47**(11):1159-1161, 2008
209. Hellemans J et al: Germline LEMD3 mutations are rare in sporadic patients with isolated melorheostosis. *Hum Mutat* **27**(3):290, 2006
210. Van Hougenhouck-Tulleken W et al: Clinical and molecular characterization of lipoid proteinosis in Namaqualand, South Africa. *Br J Dermatol* **151**(2):413-423, 2004
211. Hamada T et al: Extracellular matrix protein 1 gene (ECM1) mutations in lipoid proteinosis and genotype-phenotype correlation. *J Invest Dermatol* **120**(3):345-350, 2003
212. Sercu S et al: ECM1 interacts with fibulin-3 and the beta 3 chain of laminin 332 through its serum albumin subdomain-like 2 domain. *Matrix Biol* **28**(3):160-169, 2009
213. Fujimoto N et al: Extracellular matrix protein 1 interacts with the domain III of fibulin-1C and 1D variants through its central tandem repeat 2. *Biochem Biophys Res Commun* **333**(4):1327-1333, 2005
214. Dyer JA, Yu QC, Paller AS: "Free-floating" desmosomes in lipoid proteinosis: An inherent defect in keratinocyte adhesion? *Pediatr Dermatol* **23**(1):1-6, 2006
215. Sercu S et al: Functional redundancy of extracellular matrix protein 1 in epidermal differentiation. *Br J Dermatol* **157**(4):771-775, 2007
216. Chan I: The role of extracellular matrix protein 1 in human skin. *Clin Exp Dermatol* **29**(1):52-56, 2004
217. Kowalewski C et al: Three-dimensional imaging reveals major changes in skin microvasculature in lipoid proteinosis and lichen sclerosus. *J Dermatol Sci* **38**(3):215-224, 2005
218. Oyama N et al: Autoantibodies to extracellular matrix protein 1 in lichen sclerosus. *Lancet* **362**(9378):118-123, 2003
219. Hamada T: Lipoid proteinosis. *Clin Exp Dermatol* **27**(8):624-629, 2002
220. Sargenti NS, Batista JD, Durighetto AJ: A case of oral recurrent ulcerative lesions in a patient with lipoid proteinosis (Urbach-Wiethe disease). *Br J Oral Maxillofac Surg* **48**(8):654-655, 2009
221. Cote DN: Head and neck manifestations of lipoid proteinosis. *Otolaryngol Head Neck Surg* **119**(1):144-145, 1998

222. Siebert M, Markowitsch HJ, Bartel P: Amygdala, affect and cognition: Evidence from 10 patients with Urbach-Wiethe disease. *Brain* **126**(Pt 12):2627-2637, 2003
223. Paller AS: Histology of lipoid proteinosis. *JAMA* **272**(7):564-565, 1994
224. Ko C, Barr RJ: Vesicular lesions in a patient with lipoid proteinosis: A probable acantholytic dermatosis. *Am J Dermatopathol* **25**(4):335-337, 2003
225. Teive HA et al: Generalized dystonia and striatal calcifications with lipoid proteinosis. *Neurology* **63**(11):2168-2169, 2004
226. Chan I et al: Rapid diagnosis of lipoid proteinosis using an anti-extracellular matrix protein 1 (ECM1) antibody. *J Dermatol Sci* **35**(2):151-153, 2004
227. Ozkaya-Bayazit E et al: [Oral DMSO therapy in 3 patients with lipoidproteinosis. Results of long-term therapy]. *Hautarzt* **48**(7):477-481, 1997
228. Toosi S, Ehsani AH: Treatment of lipoid proteinosis with acitretin: A case report. *J Eur Acad Dermatol Venereol* **23**(4):482-483, 2009
229. Haneke E et al: Hyalinosis cutis et mucosae in siblings. *Hum Genet* **68**(4):342-345, 1984
230. Rosenthal G et al: Carbon dioxide laser treatment for lipoid proteinosis (Urbach-Wiethe syndrome) involving the eyelids. *Br J Ophthalmol* **81**(3):253, 1997

