

# Chapter 133

## Amyloidosis of the Skin

Helen J. Lachmann & Philip N. Hawkins

### REFERENCES

1. Pepys MB, Hawkins PN: Amyloidosis. In: *Oxford Text Book of Medicine*, 5th edition, edited by DA Warrell, TM Cox, JD Firth. Oxford University Press, 2010, pp. 1766-1779
2. Merlini G, Bellotti V: Molecular mechanisms of amyloidosis. *N Engl J Med* **349**:583-596, 2003
3. Westermark P et al: A primer of amyloid nomenclature. *Amyloid* **14**:179-183, 2007
4. Kyle RA, Gertz MA: Primary systemic amyloidosis: Clinical and laboratory features in 474 cases. *Semin Hematol* **32**:45-59, 1995
5. Comenzo RL et al: The tropism of organ involvement in primary systemic amyloidosis: Contributions of Ig V<sub>L</sub> germ line gene use and clonal plasma cell burden. *Blood* **98**:714-720, 2001
6. Lachmann HJ et al: Natural history and outcome in systemic AA amyloidosis. *N Engl J Med* **356**:2361-2371, 2007
7. Pithukpakorn M, Aksentjevich I, Toro JR: Autoinflammatory diseases: Clinical and dermatologic features, genetics, pathogenesis and therapy. *Adv Dermatol* **22**:67-90, 2006
8. Goodman HJB et al: Localized amyloidosis: Clinical features and outcome in 235 cases. *Hematologica* **90**(Suppl 1):203, 2005 [abstract]
9. Benson MD: Ostertag revisited: The inherited systemic amyloidoses without neuropathy. *Amyloid* **12**:75-87, 2005
10. Kyle RA, Bayrd ED: Amyloidosis: Review of 236 cases. *Medicine* **54**:271-299, 1975
11. Shah PL et al: The importance of complete screening for amyloid fibril type and systemic disease in patients with amyloidosis in the respiratory tract. *Sarcoidosis Vasc Diffuse Lung Dis* **19**:134-142, 2002
12. Yoshida A et al: Lichen amyloidosis induced on the upper back by long-term friction with a nylon towel. *J Dermatol* **36**(1):56-59, 2009
13. Gertz MA et al: Amyloidosis. *Best Pract Res Clin Haematol* **18**:709-727, 2005
14. Benson MD, Kincaid JC: The molecular biology and clinical features of amyloid neuropathy. *Muscle Nerve* **36**:411-423, 2007
15. Lachmann HJ, Hawkins PN: Amyloidosis, familial renal. *eMed. J. [serial online]*, 2004, [www.emedicine.com/med/topic3379.htm](http://www.emedicine.com/med/topic3379.htm)
16. Schremi S et al: Cutaneous amyloidoses and systemic amyloidoses with cutaneous involvement. *Eur J Dermatol* **20**(2):152-160, 2010
17. Ahmad QM et al: Systemic amyloidosis presenting as mucocutaneous bullous lesions. *Hematol Oncol Stem Cell Ther* **2**:418-421, 2009
18. Gillmore JD et al: Hereditary renal amyloidosis associated with variant lysozyme in a large English family. *Nephrol Dial Transplant* **14**:2639-2644, 1999
19. Rocha N et al: Cutaneous manifestations of familial amyloidotic polyneuropathy. *J Eur Acad Dermatol Venereol* **19**:605-607, 2005
20. Schwendiman MN et al: Primary cutaneous nodular amyloidosis: Case report and review of the literature. *Cutis* **84**:87-92, 2009
21. Bridoux F et al: Localized amyloidosis: Clinical features and outcome in 203 cases. *Paper presented at: Xth International Symposium on Amyloid and Amyloidosis*, 2004; Tours, France
22. Westermark P et al: Galectin 7-associated cutaneous amyloidosis. *Amyloid* **17**(Suppl 1):71, 2010
23. Meijer JM et al: Sjögren's syndrome and localized nodular cutaneous amyloidosis: Coincidence or a distinct clinical entity? *Arthritis Rheum* **58**(7):1992-1999, 2008
24. Tidman MJ, Wells RS, MacDonald DM: Pachyonychia congenita with cutaneous amyloidosis and hyperpigmentation—A distinct variant. *J Am Acad Dermatol* **16**:935-940, 1987
25. Gullu S et al: Multiple endocrine neoplasia type 2A/localized cutaneous lichen amyloidosis associated with malignant pheochromocytoma and ganglioneuroma. *J Endocrinol Invest* **28**:734-737, 2005

26. Rasi A, Khatami A, Javaheri SM: Macular amyloidosis: An assessment of prevalence, sex, and age. *Int J Dermatol* **43**:898-899, 2004
27. Wang WJ et al: Clinical and histopathological characteristics of primary cutaneous amyloidosis in 794 Chinese patients. *Zhonghua Yi Xue Za Zhi (Taipei)* **64**:101-107, 2001
28. Uenishi T et al: Hyperkeratotic variant of porokeratosis Mibelli with dermal amyloid deposits. *J Dermatol* **37**:475-479, 2010
29. Wang WJ et al: Anosacral cutaneous amyloidosis: A study of 10 Chinese cases. *Br J Dermatol* **143**:1266-1269, 2000
30. Yumlu S et al: Localized insulin-derived amyloidosis in patients with diabetes mellitus: A case report. *Hum Pathol* **40**:1655-1660, 2009
31. Lin MW et al: Novel IL31RA gene mutation and ancestral OSMR mutant allele in familial primary cutaneous amyloidosis. *Eur J Hum Genet* **18**(1):26-32, 2010
32. Gertz MA et al: Definition of organ involvement and treatment response in immunoglobulin light chain amyloidosis (AL): A consensus opinion from the 10th International Symposium on Amyloid and Amyloidosis. *Am J Hematol* **79**:319-328, 2005
33. British Committee for Standards in Haematology Working Group; Bird J et al. Guidelines on the diagnosis and management of AL amyloidosis, February 2003. *Br J Haematol* **125**:681-700, 2004
34. Katzmann JA et al: Diagnostic performance of quantitative kappa and lambda free light chain assays in clinical practice. *Clin Chem* **51**(5):878-881, 2005
35. Puchtler H, Sweat F, Levine M: On the binding of Congo red by amyloid. *J Histochem Cytochem* **10**:355-364, 1962
36. Linke RP et al: Classification of amyloidosis: Misdiagnosing by way of incomplete immunohistochemistry and how to prevent it. *Acta Histochem* **108**(3):197-208, 2006
37. Vrana JA et al: Classification of amyloidosis by laser microdissection and mass spectrometry-based proteomic analysis in clinical biopsy specimens. *Blood* **114**:4957-4959, 2009
38. Lachmann HJ et al: Misdiagnosis of hereditary amyloidosis as AL (primary) amyloidosis. *N Engl J Med* **346**:1786-1791, 2002
39. Hawkins PN, Lavender JP, Pepys MB: Evaluation of systemic amyloidosis by scintigraphy with <sup>123</sup>I-labeled serum amyloid P component. *N Engl J Med* **323**:508-513, 1990
40. Halwani O, Delgado DH: Cardiac amyloidosis: An approach to diagnosis and management. *Expert Rev Cardiovasc Ther* **8**(7):1007-1013, 2010
41. Maceira AM et al: Cardiovascular magnetic resonance and prognosis in cardiac amyloidosis. *J Cardiovasc Magn Reson* **10**:54, 2008
42. Palladini G et al: Circulating amyloidogenic free light chains and serum N-terminal natriuretic peptide type B decrease simultaneously in association with improvement of survival in AL. *Blood* **107**:3854-3858, 2006
43. Dispenzieri A et al: Serum cardiac troponins and N-terminal pro-brain natriuretic peptide: A staging system for primary systemic amyloidosis. *J Clin Oncol* **22**:3751-3757, 2004
44. Dubrey SW, Falk RH: Amyloid heart disease. *Br J Hosp Med (Lond)* **71**(2):76-82, 2010
45. Gertz MA et al: Clinical outcome of immunoglobulin light chain amyloidosis affecting the kidney. *Nephrol Dial Transplant* **24**:3132-3137, 2009
46. Sattianayagam P, Hawkins P, Gillmore J: Amyloid and the GI tract. *Expert Rev Gastroenterol Hepatol* **3**:615-630, 2009
47. Simmons Z, Specht CS: The neuromuscular manifestations of amyloidosis. *J Clin Neuromuscul Dis* **11**(3):145-157, 2010
48. Wechalekar AD, Hawkins PN, Gillmore JD: Perspectives in treatment of AL amyloidosis. *Br J Haematol* **140**:365-377, 2008
49. Herlenius G et al: Ten years of international experience with liver transplantation for familial amyloidotic polyneuropathy: Results from the familial amyloidotic polyneuropathy world transplant registry. *Transplantation* **77**:64-71, 2004
50. Gertz MA et al: Risk-adjusted manipulation of melphalan dose before stem cell transplantation in patients with amyloidosis is associated with a lower response rate. *Bone Marrow Transplant* **34**:1025-1031, 2004
51. Jaccard A et al: High-dose melphalan versus melphalan plus dexamethasone for AL amyloidosis. *N Engl J Med* **357**:1083-1093, 2007
52. Wechalekar AD et al: Safety and efficacy of risk-adapted cyclophosphamide, thalidomide, and dexamethasone in systemic AL amyloidosis. *Blood* **109**:457-464, 2007
53. Wechalekar AD et al: Efficacy of bortezomib in systemic AL amyloidosis with relapsed/refractory clonal disease. *Haematologica* **93**:295-298, 2008

54. Sanchorawala V et al: Long-term outcome of patients with AL amyloidosis treated with high-dose melphalan and stem-cell transplantation. *Blood* **110**:3561-3563, 2007
55. Moreau P et al: Prognostic factors for survival and response after high-dose therapy and autologous stem cell transplantation in systemic AL amyloidosis: A report on 21 patients. *Br J Haematol* **101**:766-769, 1998
56. Lachmann HJ, Hawkins PN: Developments in the scientific and clinical understanding of autoinflammatory disorders. *Arthritis Res Ther* **11**:212, 2009
57. Stangou AJ et al: Liver transplantation in ATTR Met30 and other variants: Time to shift the paradigm? In: *Amyloid and Amyloidosis*, edited by G Grateau, RA Kyle, M Skinner. Boca Raton, Florida: CRC Press, 2005, p. 306-308
58. Dember LM: Emerging treatment approaches for the systemic amyloidoses. *Kidney Int* **68**:1377-1390, 2005
59. Gillmore JD et al: Sustained pharmacological depletion of serum amyloid P component in patients with systemic amyloidosis. *Br J Haematol* **148**(5):760-767, 2010
60. Clos AL et al: Therapeutic removal of amyloid deposits in cutaneous amyloidosis by localised intra-lesional injections of anti-amyloid antibodies. *Exp Dermatol* **19**(10):904-911, 2010
61. Pandhi R, Kaur I, Kumar B: Lack of effect of dimethylsulphoxide in cutaneous amyloidosis. *J Dermatol Treat* **13**:11-14, 2002
62. Tran D, Kwok YK, Goh CL: A retrospective review of PUVA therapy at the National Skin Centre of Singapore. *Photodermatol Photoimmunol Photomed* **17**:164-167, 2001
63. Castanedo-Cazares JP, Lepe V, Moncada B: Lichen amyloidosis improved by 0.1% topical tacrolimus. *Dermatology* **205**:420-421, 2002
64. Grimmer J et al: Successful treatment of lichen amyloidosis with combined bath PUVA photochemotherapy and oral acitretin. *Clin Exp Dermatol* **32**(1):39-42, 2006

