

Chapter 51

Acantholytic Disorders of the Skin

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REFERENCES

1. Burge SM, Wilkinson JD: Darier-White disease: A review of the clinical features in 163 patients. *J Am Acad Dermatol* **27**:40, 1992
2. Munro CS: The phenotype of Darier's disease: Penetrance and expressivity in adults and children. *Br J Dermatol* **127**:126, 1992
3. Tavadia S, Mortimer E, Munro CS: Genetic epidemiology of Darier's disease: A population study in the west of Scotland. *Br J Dermatol* **146**:107, 2002
4. Godic A et al: Epidemiology of Darier's Disease in Slovenia. *Acta Dermatovenerol Alp Panonica Adriat* **14**:43, 2005
5. Svendsen IB, Albrectsen B: The prevalence of dyskeratosis follicularis (Darier's disease) in Denmark: An investigation of the heredity in 22 families. *Acta Derm Venereol* **39**:256, 1959
6. Craddock N et al: The gene for Darier's disease maps to chromosome 12q23-q24.1. *Hum Mol Genet* **2**:1941, 1993
7. Sakuntabhai A et al: Mutations in ATP2A2, encoding a Ca²⁺ pump, cause Darier disease. *Nat Genet* **21**:271, 1999
8. Pani B, Singh BB: Darier's disease: A calcium-signaling perspective. *Cell Mol Life Sci* **65**:205, 2008
9. Hovnanian A: SERCA pumps and human diseases. *Subcell Biochem* **45**:337, 2007
10. Hovnanian A: Darier's disease: From dyskeratosis to endoplasmic reticulum calcium ATPase deficiency. *Biochem Biophys Res Commun* **322**:1237, 2004
11. Dhitavat J et al: Calcium pumps and keratinocytes: Lessons from Darier's disease and Hailey-Hailey disease. *Br J Dermatol* **150**:821, 2004
12. Verboomen H et al: Functional difference between SERCA2a and SERCA2b Ca²⁺ pumps and their modulation by phospholamban. *Biochem J* **286**(Pt 2):591, 1992
13. Verboomen H et al: The functional importance of the extreme C-terminal tail in the gene 2 organelar Ca(2+)-transport ATPase (SERCA2a/b). *Biochem J* **303**(Pt 3):979, 1994
14. Tavadia S et al: Platelet and cardiac function in Darier's disease. *Clin Exp Dermatol* **26**:696, 2001
15. Mayosi BM et al: Heterozygous disruption of SERCA2a is not associated with impairment of cardiac performance in humans: Implications for SERCA2a as a therapeutic target in heart failure. *Heart* **92**:105, 2006
16. Tavadia S et al: Expression of the sarco/endoplasmic reticulum calcium ATPase type 2 and 3 isoforms in normal skin and Darier's disease. *Br J Dermatol* **151**:440, 2004
17. Dhitavat J et al: Mutations in the sarcoplasmic/endoplasmic reticulum Ca²⁺ ATPase isoform cause Darier's disease. *J Invest Dermatol* **121**:486, 2003
18. Ikeda S et al: Mutations in ATP2A2 in patients with Darier's disease. *J Invest Dermatol* **121**:475, 2003
19. Wuytack F, Raeymaekers L, Missiaen L: Molecular physiology of the SERCA and SPCA pumps. *Cell Calcium* **32**:279, 2002
20. Jacobsen NJ et al: ATP2A2 mutations in Darier's disease and their relationship to neuropsychiatric phenotypes. *Hum Mol Genet* **8**:1631, 1999
21. Sakuntabhai A et al: Spectrum of novel ATP2A2 mutations in patients with Darier's disease. *Hum Mol Genet* **8**:1611, 1999
22. Ruiz-Perez VL et al: ATP2A2 mutations in Darier's disease: Variant cutaneous phenotypes are associated with missense mutations, but neuropsychiatric features are independent of mutation class. *Hum Mol Genet* **8**:1621, 1999
23. Ringpfeil F et al: Darier disease—Novel mutations in ATP2A2 and genotype-phenotype correlation. *Exp Dermatol* **10**:19, 2001
24. Takahashi H et al: Novel mutations of ATP2A2 gene in Japanese patients of Darier's disease. *J Dermatol Sci* **26**:169, 2001
25. Yang Y et al: Novel point mutations of the ATP2A2 gene in two Chinese families with Darier disease. *J Invest Dermatol* **116**:482, 2001
26. Pecina-Slaus N et al: Clinical case of acral hemorrhagic Darier's disease is not caused by mutations in exon 15 of the ATP2A2 gene. *Coll Antropol* **27**:125, 2003

27. Godic A et al: P160L mutation in the Ca(2+) ATPase 2A domain in a patient with severe Darier disease. *Dermatology* **209**:142, 2004
28. Onozuka T et al: Mutational analysis of the ATP2A2 gene in two Darier disease families with intrafamilial variability. *Br J Dermatol* **150**:652, 2004
29. Racz E et al: Identification of mutations in the ATP2A2 gene in patients with Darier's disease from Hungary. *Exp Dermatol* **13**:396, 2004
30. Racz E et al: Three novel mutations in the ATP2A2 gene in Hungarian families with Darier's disease, including a novel splice site generating intronic nucleotide change. *J Dermatol Sci* **38**:231, 2005
31. Ren YQ et al: Five mutations of ATP2A2 gene in Chinese patients with Darier's disease and a literature review of 86 cases reported in China. *Arch Dermatol Res* **298**:58, 2006
32. Bchetnia M et al: Clinical and mutational heterogeneity of Darier disease in Tunisian families. *Arch Dermatol* **145**:654, 2009
33. Tsuruta D et al: Three-base deletion mutation c.120_122delGTT in ATP2A2 leads to the unique phenotype of comedonal Darier disease. *Br J Dermatol* **162**:687, 2010
34. Leinonen PT et al: Reevaluation of the normal epidermal calcium gradient, and analysis of calcium levels and ATP receptors in Hailey-Hailey and Darier epidermis. *J Invest Dermatol* **129**:1379, 2009
35. Caulfield JB, Wilgram GF: An electron-microscope study of dyskeratosis and acantholysis in Darier's disease. *J Invest Dermatol* **41**:57, 1963
36. Burge SM, Garrod DR: An immunohistological study of desmosomes in Darier's disease and Hailey-Hailey disease. *Br J Dermatol* **124**:242, 1991
37. Hashimoto K et al: Desmosomal dissolution in Grover's disease, Hailey-Hailey's disease and Darier's disease. *J Cutan Pathol* **22**:488, 1995
38. Hakuno M et al: Dissociation of intra- and extracellular domains of desmosomal cadherins and E-cadherin in Hailey-Hailey disease and Darier's disease. *Br J Dermatol* **142**:702, 2000
39. Gniadecki R et al: Relationship between keratinocyte adhesion and death: Anoikis in acantholytic diseases. *Arch Dermatol Res* **290**:528, 1998
40. Dremina ES et al: Anti-apoptotic protein Bcl-2 interacts with and destabilizes the sarcoplasmic/endoplasmic reticulum Ca2+-ATPase (SERCA). *Biochem J* **383**:361, 2004
41. Bongiorno MR, Arico M: The behaviour of Bcl-2, Bax and Bcl-x in Darier's disease. *Br J Dermatol* **147**:696, 2002
42. Pasmatzis E et al: Reduced expression of the antiapoptotic proteins of Bcl-2 gene family in the lesional epidermis of patients with Darier's disease. *J Cutan Pathol* **34**:234, 2007
43. Michalak M, Robert Parker JM, Opas M: Ca2+ signaling and calcium binding chaperones of the endoplasmic reticulum. *Cell Calcium* **32**:269, 2002
44. Kitajima Y: Mechanisms of desmosome assembly and disassembly. *Clin Exp Dermatol* **27**:684, 2002
45. Dhitavat J et al: Impaired trafficking of the desmoplakins in cultured Darier's disease keratinocytes. *J Invest Dermatol* **121**:1349, 2003
46. Burge SM et al: Darier's disease: An immunohistochemical study using monoclonal antibodies to human cytokeratins. *Br J Dermatol* **118**:629, 1988
47. Koizumi H et al: Differentiation-associated localization of small proline-rich protein in normal and diseased human skin. *Br J Dermatol* **134**:686, 1996
48. Kassab S et al: Immunohistological study of involucrin expression in Darier's disease skin. *J Cutan Pathol* **35**:635, 2008
49. Pani B et al: Up-regulation of transient receptor potential canonical 1 (TRPC1) following sarco(endo)plasmic reticulum Ca2+ ATPase 2 gene silencing promotes cell survival: A potential role for TRPC1 in Darier's disease. *Mol Biol Cell* **17**:4446, 2006
50. Liu LH et al: Squamous cell tumors in mice heterozygous for a null allele of Atp2a2, encoding the sarco(endo)plasmic reticulum Ca2+-ATPase isoform 2 Ca2+ pump. *J Biol Chem* **276**:26737, 2001
51. Hong JH et al: Markers of squamous cell carcinoma in sarco/endoplasmic reticulum Ca(2+) ATPase 2 heterozygote mice keratinocytes. *Prog Biophys Mol Biol* **103**:81-7, 2010
52. Alexandrescu DT et al: Development of squamous cell carcinomas in Darier disease: A new model for skin carcinogenesis? *Br J Dermatol* **159**:1378, 2008
53. Matsui K et al: Squamous cell carcinoma arising from Darier's disease. *Clin Exp Dermatol* **34**:e1015, 2009
54. Vazquez J et al: Vulval squamous cell carcinoma arising in localized Darier's disease. *Eur J Obstet Gynecol Reprod Biol* **102**:206, 2002

55. Orihuela E et al: Development of human papillomavirus type 16 associated squamous cell carcinoma of the scrotum in a patient with Darier's disease treated with systemic isotretinoin. *J Urol* **153**:1940, 1995
56. Downs AM, Ward KA, Peachey RD: Subungual squamous cell carcinoma in Darier's disease. *Clin Exp Dermatol* **22**:277, 1997
57. Foggia L et al: Activity of the hSPCA1 Golgi Ca²⁺ pump is essential for Ca²⁺-mediated Ca²⁺ response and cell viability in Darier disease. *J Cell Sci* **119**:671, 2006
58. Mayuzumi N et al: Effects of ultraviolet B irradiation, proinflammatory cytokines and raised extracellular calcium concentration on the expression of ATP2A2 and ATP2C1. *Br J Dermatol* **152**:697, 2005
59. Sule N et al: Lithium suppresses epidermal SERCA2 and PMR1 levels in the rat. *Pathol Oncol Res* **12**:234, 2006
60. Fong G et al: Congenital Darier disease. *J Am Acad Dermatol* **59**:S50, 2008
61. Ormerod E et al: Darier-White disease: A follow-up study. *Br J Dermatol* **161**(Suppl. 1):41, 2009
62. Burge S: Darier's disease—The clinical features and pathogenesis. *Clin Exp Dermatol* **19**:193, 1994
63. Harris A et al: Handicap in Darier's disease and Hailey-Hailey disease. *Br J Dermatol* **135**:959, 1996
64. Rallis E et al: Acrokeratosis verruciformis of Hopf (Hopf disease): Case report and review of the literature. *Dermatol Online J* **11**:10, 2005
65. Jones WN, Nix TE Jr, Clark WH Jr: Hemorrhagic Darier's Disease. *Arch Dermatol* **89**:523, 1964
66. Coulson IH, Misch KJ: Haemorrhagic Darier's disease. *J R Soc Med* **82**:365, 1989
67. Foresman PL et al: Hemorrhagic Darier's disease. *Arch Dermatol* **129**:511, 1993
68. Regazzini R et al: Isolated acral Darier's disease with haemorrhagic lesions in a kindred. *Br J Dermatol* **135**:495, 1996
69. Macleod RI, Munro CS: The incidence and distribution of oral lesions in patients with Darier's disease. *Br Dent J* **171**:133, 1991
70. Cardoso CL et al: Darier disease: Case report with oral manifestations. *Med Oral Patol Oral Cir Bucal* **11**:E404, 2006
71. Bernabe DG et al: Multiple white papules in the palate: Oral manifestation of Darier's disease. *Clin Exp Dermatol* **34**:e270, 2009
72. Vieites B et al: Darier's disease with esophageal involvement. *Scand J Gastroenterol* **43**:1020, 2008
73. Al Robaee A et al: Extensive Darier's disease with esophageal involvement. *Int J Dermatol* **43**:835, 2004
74. Klein A, Burns L, Leyden JJ: Rectal mucosa involvement in keratosis follicularis. *Arch Dermatol* **109**:560, 1974
75. Adam AE: Ectopic Darier's disease of the cervix: An extraordinary cause of an abnormal smear. *Cytopathology* **7**:414, 1996
76. Blackman HJ, Rodrigues MM, Peck GL: Corneal epithelial lesions in keratosis follicularis (Darier's disease). *Ophthalmology* **87**:931, 1980
77. Mielke J et al: Recurrent corneal ulcerations with perforation in keratosis follicularis (Darier-White disease). *Br J Ophthalmol* **86**:1192, 2002
78. Lagali N, Dellby A, Fagerholm P: In vivo confocal microscopy of the cornea in Darier-White disease. *Arch Ophthalmol* **127**:816, 2009
79. Brown VL et al: Extensive recalcitrant Darier disease successfully treated with laser ablation. *Br J Dermatol* **162**:227-229, 2010
80. Telfer NR, Burge SM, Ryan TJ: Vesiculo-bullous Darier's disease. *Br J Dermatol* **122**:831, 1990
81. Rongioletti F, Cestari R, Rebora A: Verrucous and malodorous vegetations on the legs. Darier's disease, cornifying type. *Arch Dermatol* **128**:399, 1992
82. Katta R, Reed J, Wolf JE: Cornifying Darier's disease. *Int J Dermatol* **39**:844, 2000
83. Aliagaoglu C et al: Comedonal, cornifying and hypertrophic Darier's disease in the same patient: A Darier combination. *J Dermatol* **33**:477, 2006
84. Fitzgerald DA, Lewis-Jones MS: Darier's disease presenting as isolated hyperkeratosis of the breasts. *Br J Dermatol* **136**:290, 1997
85. Derrick EK, Darley CR, Burge S: Comedonal Darier's disease. *Br J Dermatol* **132**:453, 1995
86. Lee MW et al: Two cases of comedonal Darier's disease. *Clin Exp Dermatol* **27**:714, 2002
87. Millard TP et al: 'Groveroid' Darier's disease? *Br J Dermatol* **150**:600, 2004
88. Ohtake N et al: Brown papules and leukoderma in Darier's disease: Clinical and histological features. *Dermatology* **188**:157, 1994
89. Goh BK, Ang P, Goh CL: Darier's disease in Singapore. *Br J Dermatol* **152**:284, 2005

90. Munro CS, Cox NH: An acantholytic dyskeratotic epidermal naevus with other features of Darier's disease on the same side of the body. *Br J Dermatol* **127**:168, 1992
91. Reese DA, Paul AY, Davis B: Unilateral segmental Darier disease following Blaschko lines: A case report and review of the literature. *Cutis* **76**:197, 2005
92. Sakuntabhai A et al: Mosaicism for ATP2A2 mutations causes segmental Darier's disease. *J Invest Dermatol* **115**:1144, 2000
93. Wada T et al: A Japanese case of segmental Darier's disease caused by mosaicism for the ATP2A2 mutation. *Br J Dermatol* **149**:185, 2003
94. Gilaberte M et al: Acantholytic dyskeratotic naevi following Blaschko's lines: A mosaic form of Darier's disease. *J Eur Acad Dermatol Venereol* **17**:196, 2003
95. Happle R, Itin PH, Brun AM: Type 2 segmental Darier disease. *Eur J Dermatol* **9**:449, 1999
96. Ehrh U, Brieger P: Comorbidity of keratosis follicularis (Darier's disease) and bipolar affective disorder: An indication for valproate instead of lithium. *Gen Hosp Psychiatry* **22**:128, 2000
97. Wang SL et al: Darier's disease associated with bipolar affective disorder: A case report. *Kaohsiung J Med Sci* **18**:622, 2002
98. Gordon-Smith K et al: The neuropsychiatric phenotype in Darier disease. *Br J Dermatol* **161**(Suppl.1):40, 2009
99. Clark RD Jr, Hammer CJ, Patterson SD: A cutaneous disorder (Darier's disease) evidently exacerbated by lithium carbonate. *Psychosomatics* **27**:800, 1986
100. Milton GP et al: Exacerbation of Darier's disease by lithium carbonate. *J Am Acad Dermatol* **23**:926, 1990
101. Rubin MB: Lithium-induced Darier's disease. *J Am Acad Dermatol* **32**:674, 1995
102. Denicoff KD et al: Suicidal ideation in Darier's disease. *J Am Acad Dermatol* **22**:196, 1990
103. Jones I et al: Evidence for familial cosegregation of major affective disorder and genetic markers flanking the gene for Darier's disease. *Mol Psychiatry* **7**:424, 2002
104. Jacobsen NJ et al: Exclusion of the Darier's disease gene, ATP2A2, as a common susceptibility gene for bipolar disorder. *Mol Psychiatry* **6**:92, 2001
105. Green EK et al: P2RX7: A bipolar and unipolar disorder candidate susceptibility gene? *Am J Med Genet B Neuropsychiatr Genet* **150B**:1063-1069, 2009
106. Ramien ML et al: Cystic bone lesions in a boy with Darier disease: A magnetic resonance imaging assessment. *J Am Acad Dermatol* **60**:1062, 2009
107. Castori M et al: Darier disease, multiple bone cysts, and aniridia due to double de novo heterozygous mutations in ATP2A2 and PAX6. *Am J Med Genet A* **149A**:1768, 2009
108. Crisp AJ et al: The prevalence of bone cysts in Darier's disease: A survey of 31 cases. *Clin Exp Dermatol* **9**:78, 1984
109. Hall JR et al: Familial dyskeratotic comedones. A report of three cases and review of the literature. *J Am Acad Dermatol* **17**:808, 1987
110. Hallermann C, Bertsch HP: Two sisters with familial dyskeratotic comedones. *Eur J Dermatol* **14**:214, 2004
111. Nakagawa T et al: Comedo-like acantholytic dyskeratosis of the face and scalp: A new entity? *Br J Dermatol* **142**:1047, 2000
112. Burge SM: Hailey-Hailey disease: The clinical features, response to treatment and prognosis. *Br J Dermatol* **126**:275, 1992
113. Gilchrist H et al: Galli-Galli disease: A case report with review of the literature. *J Am Acad Dermatol* **58**:299, 2008
114. Mittag H, Rupec M, Klingmuller G: Galli-Galli's disease: An entity? A clinical, histological and electron microscopic study. *Aktuelle Dermatologie* **12**:41, 1986
115. Niedleman ML, Mc KV: Acrokeratosis verruciformis (Hopf). A follow-up study. *Arch Dermatol* **86**:779, 1962
116. Dhitavat J et al: Acrokeratosis verruciformis of Hopf is caused by mutation in ATP2A2: Evidence that it is allelic to Darier's disease. *J Invest Dermatol* **120**:229, 2003
117. Browne F et al: Papular acantholytic dyskeratosis presenting as genital warts. *Int J STD AIDS* **18**:867, 2007
118. Salopek TG, Krol A, Jimbow K: Case report of Darier disease localized to the vulva in a 5-year-old girl. *Pediatr Dermatol* **10**:146, 1993
119. Langenberg A et al: Genital benign chronic pemphigus (Hailey-Hailey disease) presenting as condylomas. *J Am Acad Dermatol* **26**:951, 1992
120. Cooper PH: Acantholytic dermatosis localized to the vulvocrural area. *J Cutan Pathol* **16**:81, 1989

121. Parham DM et al: Disseminated herpes simplex infection complicating Darier's disease: Successful treatment with oral acyclovir. *J Infect* **10**:77, 1985
122. Kandasamy R et al: Darier disease complicated by disseminated zoster. *Dermatol Online J* **15**:6, 2009
123. Tegner E, Jonsson N: Darier's disease with involvement of both submandibular glands. *Acta Derm Venereol* **70**:451, 1990
124. Micali G, Nasca MR: Tazarotene gel in childhood Darier disease. *Pediatr Dermatol* **16**:243, 1999
125. Brazzelli V et al: Linear Darier's disease successfully treated with 0.1% tazarotene gel "short-contact" therapy. *Eur J Dermatol* **16**:59, 2006
126. Burge SM, Buxton PK: Topical isotretinoin in Darier's disease. *Br J Dermatol* **133**:924, 1995
127. Casals M et al: Successful treatment of linear Darier's disease with topical adapalene. *J Eur Acad Dermatol Venereol* **23**:237, 2009
128. Cianchini G et al: Acral Darier's disease successfully treated with adapalene. *Acta Derm Venereol* **81**:57, 2001
129. Knulst AC, De La Faille HB, Van Vloten WA: Topical 5-fluorouracil in the treatment of Darier's disease. *Br J Dermatol* **133**:463, 1995
130. Schmidt H et al: Topical 5-fluorouracil in Darier disease. *Br J Dermatol* **158**:1393, 2008
131. Yoon TY, Kim JW, Kim MK: Successful treatment of Darier disease with topical 5-fluorouracil. *Br J Dermatol* **154**:1210, 2006
132. Rubegni P et al: A case of Darier's disease successfully treated with topical tacrolimus. *J Eur Acad Dermatol Venereol* **20**:84, 2006
133. Cooper SM, Burge SM: Darier's disease: Epidemiology, pathophysiology, and management. *Am J Clin Dermatol* **4**:97, 2003
134. Shahidullah H, Humphreys F, Beveridge GW: Darier's disease: Severe eczematization successfully treated with cyclosporin. *Br J Dermatol* **131**:713, 1994
135. Stewart LC, Yell J: Vulval Darier's disease treated successfully with ciclosporin. *J Obstet Gynaecol* **28**:108, 2008
136. Exadaktylou D et al: Treatment of Darier's disease with photodynamic therapy. *Br J Dermatol* **149**:606, 2003
137. Minsue Chen T, Wanitphakdeedecha R, Nguyen TH: Carbon dioxide laser ablation and adjunctive destruction for Darier-White disease (keratosis follicularis). *Dermatol Surg* **34**:1431, 2008
138. Santiago-et-Sanchez-Mateos JL et al: Botulinum toxin type A for the preventive treatment of intertrigo in a patient with Darier's disease and inguinal hyperhidrosis. *Dermatol Surg* **34**:1733, 2008
139. Cohen PR: Darier disease: Sustained improvement following reduction mammoplasty. *Cutis* **72**:124, 2003
140. Wang PG et al: Genetic heterogeneity in acrokeratosis verruciformis of Hopf. *Clin Exp Dermatol* **31**:558, 2006
141. Bukhari I: Acrokeratosis Verruciformis of Hopf: A localized variant. *J Drugs Dermatol* **3**:687, 2004
142. Serarslan G, Balci DD, Homan S: Acitretin treatment in acrokeratosis verruciformis of Hopf. *J Dermatolog Treat* **18**:123, 2007
143. Hu Z et al: Mutations in ATP2C1, encoding a calcium pump, cause Hailey-Hailey disease. *Nat Genet* **24**:61, 2000
144. Sudbrak R et al: Hailey-Hailey disease is caused by mutations in ATP2C1 encoding a novel Ca(2+) pump. *Hum Mol Genet* **9**:1131, 2000
145. Missiaen L et al: Calcium in the Golgi apparatus. *Cell Calcium* **41**:405, 2007
146. Vanoevelen J et al: Diseases involving the Golgi calcium pump. *Subcell Biochem* **45**:385, 2007
147. Missiaen L et al: SPCA1 pumps and Hailey-Hailey disease. *Biochem Biophys Res Commun* **322**:1204, 2004
148. Ikeda S et al: Mutations of ATP2C1 in Japanese patients with Hailey-Hailey disease: Intrafamilial and interfamilial phenotype variations and lack of correlation with mutation patterns. *J Invest Dermatol* **117**:1654, 2001
149. Dobson-Stone C et al: Hailey-Hailey disease: Molecular and clinical characterization of novel mutations in the ATP2C1 gene. *J Invest Dermatol* **118**:338, 2002
150. Chao SC, Tsai YM, Yang MH: Mutation analysis of ATP2C1 gene in Taiwanese patients with Hailey-Hailey disease. *Br J Dermatol* **146**:595, 2002
151. Yokota K et al: Analysis of ATP2C1 gene mutation in 10 unrelated Japanese families with Hailey-Hailey disease. *J Invest Dermatol* **118**:550, 2002
152. Li H, Sun XK, Zhu XJ: Four novel mutations in ATP2C1 found in Chinese patients with Hailey-Hailey disease. *Br J Dermatol* **149**:471, 2003
153. Fairclough RJ et al: Hailey-Hailey disease: Identification of novel mutations in ATP2C1 and effect of missense mutation A528P on protein expression levels. *J Invest Dermatol* **123**:67, 2004

154. Majore S et al: ATP2C1 gene mutation analysis in Italian patients with Hailey-Hailey disease. *J Invest Dermatol* 125:933, 2005
155. Ohtsuka T et al: Novel mutation in ATP2C1 gene in a Japanese patient with Hailey-Hailey disease. *Dermatology* 212:194, 2006
156. Racz E, Csikos M, Karpati S: Novel mutations in the ATP2C1 gene in two patients with Hailey-Hailey disease. *Clin Exp Dermatol* 30:575, 2005
157. Zhang XQ et al: Mutations in the ATP2C1 gene in Chinese patients with Hailey-Hailey disease. *Clin Exp Dermatol* 31:702, 2006
158. Li X et al: Two novel mutations of the ATP2C1 gene in Chinese patients with Hailey-Hailey disease. *Arch Dermatol Res* 299:209, 2007
159. Nemoto-Hasebe I et al: Diagnosis of Hailey-Hailey disease facilitated by DNA testing: A novel mutation in ATP2C1. *Acta Derm Venereol* 88:399, 2008
160. Hamada T et al: Molecular and clinical characterization in Japanese and Korean patients with Hailey-Hailey disease: Six new mutations in the ATP2C1 gene. *J Dermatol Sci* 51:31, 2008
161. Cialfi S et al: Complex multipathways alterations and oxidative stress are associated with Hailey-Hailey disease. *Br J Dermatol* 162:518, 2010
162. Fairclough RJ et al: Effect of Hailey-Hailey Disease mutations on the function of a new variant of human secretory pathway Ca²⁺/Mn²⁺-ATPase (hSPCA1). *J Biol Chem* 278:24721, 2003
163. Behne MJ et al: Human keratinocyte ATP2C1 localizes to the Golgi and controls Golgi Ca²⁺ stores. *J Invest Dermatol* 121:688, 2003
164. Ton VK, Rao R: Expression of Hailey-Hailey disease mutations in yeast. *J Invest Dermatol* 123:1192, 2004
165. Rice WJ, MacLennan DH: Scanning mutagenesis reveals a similar pattern of mutation sensitivity in transmembrane sequences M4, M5 and M6, but not in M8, of the Ca²⁺-ATPase of sarcoplasmic reticulum (SERCA1a). *J Biol Chem* 271:31412, 1996
166. Wilgram G, Caulfield J, Lever W: An electron microscopic study of acantholysis and dyskeratosis in Hailey and Hailey's disease. *J Invest Dermatol* 39:373, 1962
167. Gottlieb SK, Lutzner MA: Hailey-Hailey disease: An electron microscopic study. *J Invest Dermatol* 54:368, 1970
168. Inohara S et al: Immunohistochemical localization of desmosomal and cytoskeletal proteins in the epidermis of healthy individuals and patients with Hailey-Hailey's disease. *Acta Derm Venereol* 70:239, 1990
169. Bergman R et al: A study of keratin expression in benign familial chronic pemphigus. *Am J Dermatopathol* 14:32, 1992
170. Burge SM, Schomberg KH: Adhesion molecules and related proteins in Darier's disease and Hailey-Hailey disease. *Br J Dermatol* 127:335, 1992
171. Harada M, Hashimoto K, Fujiwara K: Immunohistochemical distribution of CD44 and desmoplakin I & II in Hailey-Hailey's disease and Darier's disease. *J Dermatol* 21:389, 1994
172. Haftek M et al: Internalization of gap junctions in benign familial pemphigus (Hailey-Hailey disease) and keratosis follicularis (Darier's disease). *Br J Dermatol* 141:224, 1999
173. Burge SM, Millard PR, Wojnarowska F: Hailey-Hailey disease: A widespread abnormality of cell adhesion. *Br J Dermatol* 124:329, 1991
174. Yoshida M et al: ATP2C1 is specifically localized in the basal layer of normal epidermis and its depletion triggers keratinocyte differentiation. *J Dermatol Sci* 43:21, 2006
175. Kawada H et al: Transcriptional regulation of ATP2C1 gene by Sp1 and YY1 and reduced function of its promoter in Hailey-Hailey disease keratinocytes. *J Invest Dermatol* 124:1206, 2005
176. Okunade GW et al: Loss of the Atp2c1 secretory pathway Ca²⁺-ATPase (SPCA1) in mice causes golgi stress, apoptosis, and midgestational death in homozygous embryos and squamous cell tumors in adult heterozygotes. *J Biol Chem* 282:26517, 2007
177. Callewaert G et al: Similar Ca(2+)-signaling properties in keratinocytes and in COS-1 cells overexpressing the secretory-pathway Ca(2+)-ATPase SPCA1. *Cell Calcium* 34:157, 2003
178. Melino G et al: Itch: A HECT-type E3 ligase regulating immunity, skin and cancer. *Cell Death Differ* 15:1103, 2008
179. Leinonen PT et al: Keratinocytes cultured from patients with Hailey-Hailey disease and Darier disease display distinct patterns of calcium regulation. *Br J Dermatol* 153:113, 2005
180. Aberg KM et al: Involucrin expression is decreased in Hailey-Hailey keratinocytes owing to increased involucrin mRNA degradation. *J Invest Dermatol* 127:1973, 2007
181. Aronchik I et al: Actin reorganization is abnormal and cellular ATP is decreased in Hailey-Hailey keratinocytes. *J Invest Dermatol* 121:681, 2003
182. Kirtschig G, Effendy I, Happle R: Leukonychia longitudinalis: als ein leitsymptom des morbus Hailey-Hailey. *Hautarzt* 43:451, 1992

183. Meawad OB, Assaf HM: Longitudinal white streaks of fingernails: A useful clinical marker in genital verruroid Hailey-Hailey disease. *J Eur Acad Dermatol Venereol* 5:177, 1995
184. Marsch WC, Stuttgen G: Generalized Hailey-Hailey disease. *Br J Dermatol* 99:553, 1978
185. Hahn H et al: Generalisierter pemphigus chronicus benignus familiaris (morbus Hailey-Hailey). *Aktuelle Dermatologie* 16:80, 1990
186. Meffert JJ, Davis BM, Campbell JC: Bullous drug eruption to griseofulvin in a man with Hailey-Hailey disease. *Cutis* 56:279, 1995
187. Gisondi P et al: Severe impairment of quality of life in Hailey-Hailey disease. *Acta Dermato-Venereologica* 85:132, 2005
188. Galimberti RL et al: Chronic benign familial pemphigus. *Int J Dermatol* 27:495, 1988
189. Richard G, Linse R, Harth W: Morbus Hailey-Hailey. Fruherfassung von merkmalsstragern durch einen UV-provokationstest-klinische relevanz der methode UV. *Hautarzt* 44:376, 1993
190. Marren P, Burge S: Seborrhoeic dermatitis of the scalp—A manifestation of Hailey-Hailey disease in a predisposed individual? *Br J Dermatol* 126:294, 1992
191. Odia SG et al: Photoprovokation eines morbus Hailey-Hailey mit konsekutivem nachweis zirkulierender pemphigusantikörper. *Aktuelle Dermatologie* 21:136, 1995
192. Ponyai G et al: Benign familial chronic pemphigus (Hailey-Hailey) provoked by contact sensitivity in 2 patients. *Contact Dermatitis* 40:168, 1999
193. Peppiatt T, Keefe M, White JE: Hailey-Hailey disease—Exacerbation by herpes simplex virus and patch tests. *Clin Exp Dermatol* 17:201, 1992
194. Rudolph CM et al: Contact irritation provoking Hailey-Hailey disease. *Contact Dermatitis* 44:371, 2001
195. Gerdson R et al: Hailey-Hailey disease: Exacerbation by scabies. *Br J Dermatol* 144:211, 2001
196. Walker SL, Beck MH: Undiagnosed Hailey-Hailey disease causing painful erosive skin changes during patch testing. *Br J Dermatol* 153:233, 2005
197. Fischer H, Nikolowski W: Die mundschleimhaut beim pemphigus benignus familiaris chronicus. *Arch Klin Exp Dermatol* 214:261, 1962
198. Schneider W, Fischer H, Wiehl R: Zur frage der schleimhautbeteiligung beim pemphigus benignus familiaris chronicus. *Arch Klin Exp Dermatol* 225:74, 1966
199. Botvinick I: Familial benign pemphigus with oral mucous membrane lesions. *Cutis* 12:371, 1973
200. Kahn D, Hutchinson E: Esophageal involvement in familial benign chronic pemphigus. *Arch Dermatol* 109:718, 1974
201. Heinze VR: Pemphigus chronicus benignus familiaris (Gougerot-Hailey-Hailey) mit schleimhautbeteiligung bei einer diabetikerin. *Dermatol Monatsschr* 165:862, 1979
202. Václavíková V, Neumann E: Vaginal involvement in familial benign chronic pemphigus (Morbus Hailey-Hailey). *Acta Dermato-Venereologica* 62:80, 1982
203. Oguz O et al: Conjunctival involvement in familial chronic benign pemphigus (Hailey-Hailey disease). *Int J Dermatol* 36:282, 1997
204. Hwang LY et al: Type 1 segmental manifestation of Hailey-Hailey disease. *J Am Acad Dermatol* 49:712, 2003
205. Poblete-Gutierrez P et al: Allelic loss underlies type 2 segmental Hailey-Hailey disease, providing molecular confirmation of a novel genetic concept. *J Clin Invest* 114:1467, 2004
206. Metzke D et al: Involvement of the adherens junction-actin filament system in acantholytic dyskeratosis of Hailey-Hailey disease. A histological, ultrastructural, and histochemical study of lesional and non-lesional skin. *J Cutan Pathol* 23:211, 1996
207. Chave TA, Milligan A: Acute generalized Hailey-Hailey disease. *Clin Exp Dermatol* 27:290, 2002
208. Ewald K, Gross G: Perianal Hailey-Hailey disease: An unusual differential diagnosis of condylomata acuminata. *Int J STD AIDS* 19:791, 2008
209. Lipoff JB et al: Acantholytic dermatosis of the crural folds with ATP2C1 mutation is a possible variant of Hailey-Hailey Disease. *J Cutan Med Surg* 13:151, 2009
210. Evron S, Leviatan A, Okon E: Familial benign chronic pemphigus appearing as leukoplakia of the vulva. *Int J Dermatol* 23:556, 1984
211. Wong TY, Mihm Mc Jr. Acantholytic dermatosis localized to genitalia and crural areas of male patients: A report of three cases. *J Cutan Pathol* 21:27, 1994
212. Dittmer CJ et al: Successful laser therapy of a papular acantholytic dyskeratosis of the vulva: Case report and review of literature. *Arch Gynecol Obstet* 281:723-725, 2010
213. Mashiko M et al: Bacterial infection-induced generalized Hailey-Hailey disease successfully treated by tretinate. *Clin Exp Dermatol* 31:57, 2006

214. Otsuka F et al: Generalized herpes simplex. Complicating Hailey-Hailey's disease. *J Dermatol* **8**:63, 1981
215. Schirren H et al: Exazerbation eines morbus Hailey-Hailey durch infektion mit Herpes simplex virus. Nachweis mittels polymerasekettenreaktion. *Hautarzt* **46**:494, 1995
216. Stallmann D, Schmoeckel C: Morbus Hailey-Hailey mit dissemination und eczema herpeticatum unter etretinattherapie. *Hautarzt* **39**:454, 1988
217. Mak RK et al: Hailey-Hailey disease failing to respond to treatment. *Clin Exp Dermatol* **30**:598, 2005
218. Reitamo S et al: Contact allergies in patients with familial benign chronic pemphigus (Hailey-Hailey disease). *J Am Acad Dermatol* **21**:506, 1989
219. Remitz A et al: Darier's disease, familial benign chronic pemphigus (Hailey-Hailey disease) and contact hypersensitivity. *J Am Acad Dermatol* **22**:134, 1990
220. Furue M et al: Basal cell epithelioma arising in a patient with Hailey-Hailey's disease. *Int J Dermatol* **26**:461, 1987
221. Ochiai T et al: Human papillomavirus types 16 and 39 in a vulvar carcinoma occurring in a woman with Hailey-Hailey disease. *Br J Dermatol* **140**:509, 1999
222. Holst VA et al: Squamous cell carcinoma arising in Hailey-Hailey disease. *J Am Acad Dermatol* **43**:368, 2000
223. Chen MY et al: Presence of human papillomavirus type 6 DNA in the perineal verrucoid lesions of Hailey-Hailey disease. *J Eur Acad Dermatol Venereol* **20**:1356, 2006
224. Chan CC et al: Human papillomavirus type 5 infection in a patient with Hailey-Hailey disease successfully treated with imiquimod. *Br J Dermatol* **156**:579, 2007
225. Cockayne SE, Rassl DM, Thomas SE: Squamous cell carcinoma arising in Hailey-Hailey disease of the vulva. *Br J Dermatol* **142**:540, 2000
226. Korner J et al: Familial cosegregation of affective disorder and Hailey-Hailey disease. *Br J Psychiatry* **163**:109, 1993
227. Wilk M et al: Pemphigus chronicus benignus familiaris (morbus Hailey-Hailey) und biplare affektive erkrankung bei drei mitgliedern einer familie. *Hautarzt* **45**:313, 1994
228. Yokota K, Sawamura D: Hailey-Hailey disease with affective disorder: Report of a case with novel ATP2C1 gene mutation. *J Dermatol Sci* **43**:150, 2006
229. Kellermayer R et al: Aminoglycosides as potential pharmacogenetic agents in the treatment of Hailey-Hailey disease. *J Invest Dermatol* **126**:229, 2006
230. Umar SA, Bhattacharjee P, Brodell RT: Treatment of Hailey-Hailey disease with tacrolimus ointment and clobetasol propionate foam. *J Drugs Dermatol* **3**:200, 2004
231. Bianchi L, Chimenti MS, Giunta A: Treatment of Hailey-Hailey disease with topical calcitriol. *J Am Acad Dermatol* **51**:475, 2004
232. Rajpara SM, King CM: Hailey-Hailey disease responsive to topical calcitriol. *Br J Dermatol* **152**:816, 2005
233. Aoki T et al: 1alpha,24-dihydroxyvitamin D3 (tacalcitol) is effective against Hailey-Hailey disease both in vivo and in vitro. *Br J Dermatol* **139**:897, 1998
234. Dammak A et al: Successful treatment of Hailey-Hailey disease with topical 5-fluorouracil. *Br J Dermatol* **161**:967, 2009
235. Rabeni EJ, Cunningham NM: Effective treatment of Hailey-Hailey disease with topical tacrolimus. *J Am Acad Dermatol* **47**:797, 2002
236. Reuter J, Termeer C, Bruckner-Tuderman L: [Tacrolimus—a new therapeutic option for Hailey-Hailey-disease?]. [Article in German.] *J Dtsch Dermatol Ges* **3**:278, 2005
237. Sand C, Thomsen HK: Topical tacrolimus ointment is an effective therapy for Hailey-Hailey disease. *Arch Dermatol* **139**:1401, 2003
238. Persic-Vojinovic S et al: Disseminated Hailey-Hailey disease treated with topical tacrolimus and oral erythromycin: Case report and review of the literature. *Acta Dermatovenerol Croat* **14**:253, 2006
239. Laffitte E, Panizzon RG: Is topical tacrolimus really an effective therapy for Hailey-Hailey disease? *Arch Dermatol* **140**:1282, 2004
240. Defresne C, Adam C, De Marneffe K: Pemphigus chronique benin familial de Hailey-Hailey. *Dermatologica* **165**:624, 1982
241. Delfino M et al: Il pemfigo familiare benigno di Hailey-Hailey terapia con calcitriolo. *Ann Ital Dermatolog Clin Sper* **44**:337, 1990
242. Hunt MJ et al: Vesiculobullous Hailey-Hailey disease: Successful treatment with oral retinoids. *Australas J Dermatol* **37**:196, 1996
243. Berger EM, Galadari HI, Gottlieb AB: Successful treatment of Hailey-Hailey disease with acitretin. *J Drugs Dermatol* **6**:734, 2007

244. Vilarinho C, Ventura F, Brito C: Methotrexate for refractory Hailey-Hailey disease. *J Eur Acad Dermatol Venereol* **24**:106, 2010
245. Cecchi R et al: Pemfigo familiare benigno (malattia di Hailey-Hailey) trattato con ciclosporina A orale. *G Ital Dermatol Venereol* **128**:615, 1993
246. BerthJones J, Smith SG, GrahamBrown RAC: Benign familial chronic pemphigus (Hailey-Hailey disease) responds to cyclosporin. *Clin Expt Dermatol* **20**:70, 1995
247. Sire D, Johnson B: Benign familial chronic pemphigus treated with Dapsone. *Arch Dermatol* **103**:262, 1971
248. Tomecki K: Hailey-Hailey disease: Clinical patterns and response to dapsone. *Clin Res* **30**:719A, 1982
249. Norman R, Greenberg RG, Jackson JM: Case reports of etanercept in inflammatory dermatoses. *J Am Acad Dermatol* **54**:S139, 2006
250. Hurd DS, Johnston C, Bevins A: A case report of Hailey-Hailey disease treated with alefacept (Amevive). *Br J Dermatol* **158**:399, 2008
251. Shons AR: Wide excision of perineal Hailey-Hailey disease with healing by secondary intention. *Br J Plast Surg* **42**:230, 1989
252. Menz P, Jackson IT, Connolly S: Surgical control of Hailey-Hailey disease. *Br J Plast Surg* **40**:557, 1987
253. Guerin-Surville H et al: Traitement chirurgical de la maladie de Hailey-Hailey par greffes chirurgicales. (2e presentation): resultats avec 5 ans de recul. *Ann Dermatol Venereol* **116**:904, 1989
254. Hamm H, Metz D, Brocker EB: Hailey-Hailey disease: Eradication by dermabrasion. *Arch Dermatol* **130**:1143, 1994
255. Kirtschig G, Gieler U, Happle R: Treatment of Hailey-Hailey disease by dermabrasion. *J Am Acad Dermatol* **28**:784, 1993
256. Konrad H, Karamfilov T, Wollina U: Intracutaneous botulinum toxin A versus ablative therapy of Hailey-Hailey disease—A case report. *J Cosmet Laser Ther* **3**:181, 2001
257. Kartamaa M, Reitamo S: Familial benign chronic pemphigus (Hailey-Hailey disease). Treatment with carbon dioxide laser vaporization. *Arch Dermatol* **128**:646, 1992
258. Kruppa A et al: Successful treatment of Hailey-Hailey disease with a scanned carbon dioxide laser. *Acta Derm Venereol* **80**:53, 2000
259. McElroy JA, Mehregan DA, Roenigk RK: Carbon dioxide laser vaporization of recalcitrant symptomatic plaques of Hailey-Hailey disease and Darier's disease. *J Am Acad Dermatol* **23**:893, 1990
260. Roos DE, Reid CM: Benign familial pemphigus: Little benefit from superficial radiotherapy. *Australas J Dermatol* **43**:305, 2002
261. Narbutt J et al: Effective treatment of recalcitrant Hailey-Hailey disease with electron beam radiotherapy. *J Eur Acad Dermatol Venereol* **21**:567, 2007
262. Ruiz-Rodriguez R et al: Photodynamic therapy with 5-aminolevulinic acid for recalcitrant familial benign pemphigus (Hailey-Hailey disease). *J Am Acad Dermatol* **47**:740, 2002
263. Fernandez Guarino M et al: Experience with photodynamic therapy in Hailey-Hailey disease. *J Dermatolog Treat* **19**:288, 2008
264. Koeysers WJ, Van Der Geer S, Krekels G: Botulinum toxin type A as an adjuvant treatment modality for extensive Hailey-Hailey disease. *J Dermatolog Treat* **19**:251, 2008
265. Lapiere JC et al: Botulinum toxin type A for the treatment of axillary Hailey-Hailey disease. *Dermatol Surg* **26**:371, 2000
266. Grover RW: Transient acantholytic dermatosis. *Arch Dermatol* **101**:426, 1970
267. Chalet M, Grover R, Ackerman AB: Transient acantholytic dermatosis: A reevaluation. *Arch Dermatol* **113**:431, 1977
268. Heenan PJ, Quirk CJ: Transient acantholytic dermatosis. *Br J Dermatol* **102**:515, 1980
269. Parsons JM: Transient acantholytic dermatosis (Grover's disease): A global perspective. *J Am Acad Dermatol* **35**:653, 1996
270. Powell J et al: Grover's disease, despite histological similarity to Darier's disease, does not share an abnormality in the ATP2A2 gene. *Br J Dermatol* **143**:658, 2000
271. Cooper SM et al: Extensive Grover's-like eruption with lentiginous 'freckling': Report of two cases. *Br J Dermatol* **150**:350, 2004
272. Hu CH, Michel B, Farber EM: Transient acantholytic dermatosis (Grover's disease). A skin disorder related to heat and sweating. *Arch Dermatol* **121**:1439, 1985
273. French LE et al: Incidence of transient acantholytic dermatosis (Grover's disease) in a hospital setting. *Dermatology* **198**:410, 1999
274. Fujita Y, Sato-Matsumura KC, Ohnishi K: Transient acantholytic dermatosis associated with B symptoms of follicular lymphoma. *Clin Exp Dermatol* **32**:752, 2007
275. Guana AL, Cohen PR: Transient acantholytic dermatosis in oncology patients. *J Clin Oncol* **12**:1703, 1994

276. Scheinfeld N, Mones J: Seasonal variation of transient acantholytic dyskeratosis (Grover's disease). *J Am Acad Dermatol* **55**:263, 2006
277. Grover RW, Rosenbaum R: The association of transient acantholytic dermatosis with other skin diseases. *J Am Acad Dermatol* **11**:253, 1984
278. Davis MD et al: Grover's disease: Clinicopathologic review of 72 cases. *Mayo Clin Proc* **74**:229, 1999
279. Rosina P et al: Grover's disease (transient acantholytic dermatosis) associated with atopy. *J Eur Acad Dermatol Venereol* **19**:390, 2005
280. Ishibashi M, Nagasaka T, Chen KR: Remission of transient acantholytic dermatosis after the treatment with rituximab for follicular lymphoma. *Clin Exp Dermatol* **33**:206, 2008
281. Held JL, Bank D, Grossman ME: Grover's disease provoked by ionizing radiation. *J Am Acad Dermatol* **19**:137, 1988
282. Tschärner GG et al: Grover's disease induced by cetuximab. *Dermatology* **213**:37, 2006
283. Boutli F et al: Transient acantholytic dermatosis (Grover's disease) in a renal transplant patient. *J Dermatol* **33**:178, 2006
284. Maghraoui S et al: Dermatose acantholytique transitoire (maladie de Grover). *Ann Dermatol Venereol* **122**:801, 1995
285. Streit M et al: Transitorische akantolytische Dermatose (M. Grover). Eine Analyse des klinischen Spektrums anhand von 21 histologisch erfassten Fällen. *Hautarzt* **51**:244, 2000
286. Waisman M, Stewart JJ, Walker AE: Bullous transient acantholytic dermatosis. *Arch Dermatol* **112**:1440, 1976
287. Quirk CJ, Heenan PJ: Grover's disease: 34 years on. *Australas J Dermatol* **45**:83, 2004
288. Girard C et al: Persistent acantholytic dermatosis and extensive lentiginous 'freckling': A new entity? *Br J Dermatol* **153**:217, 2005
289. Rowley MJ et al: Hypopigmented macules in acantholytic disorders. *Int J Dermatol* **34**:390, 1995
290. Fantini F, Kovacs E, Scarabello A: Unilateral transient acantholytic dermatosis (Grover's disease) along Blaschko lines. *J Am Acad Dermatol* **47**:319, 2002
291. Garçon N et al: Paraneoplastic transient acantholytic dermatosis (Grover's disease) along Blaschko lines. *Eur J Dermatol* **19**:405, 2009
292. Simon RS, Bloom D, Ackerman AB: Persistent acantholytic dermatosis. A variant of transient acantholytic dermatosis (Grover disease). *Arch Dermatol* **112**:1429, 1976
293. Fawcett HA, Miller JA: Persistent acantholytic dermatosis related to actinic damage. *Br J Dermatol* **109**:349, 1983
294. Eros N, Kovacs A, Karolyi Z: Successful treatment of transient acantholytic dermatosis with systemic steroids. *J Dermatol* **25**:469, 1998
295. Keohane SG, Cork MJ: Treatment of Grover's disease with calcipotriol (Dovonex). *Br J Dermatol* **132**:832, 1995
296. Mota AV et al: Successful treatment of Grover's disease with calcipotriol. *Eur J Dermatol* **8**:33, 1998
297. Hayashi H et al: Treatment of Grover's disease with tacalcitol. *Clin Exp Dermatol* **27**:160, 2002
298. Miljkovic J, Marko PB: Grover's disease: Successful treatment with acitretin and calcipotriol. *Wien Klin Wochenschr* **116**(Suppl. 2):81, 2004