Conflict of interest
The authors declare no conflict of interest.

References

1 Yooh HS, Brooks Jr HL, Capone Jr A, L'Hernault NL, Grossniklaus HE. Ultrastructural features of tissue removed during idiopathic macular hole surgery. *Am J Ophthalmol* 1996; **122**: 67–75.
2 Eckardt C, Eckardt U, Groos S, Luciano L, Reale E. Removal of the internal limiting membrane in macular holes. Clinical and morphological findings. *Ophthalmologe* 1997; **94**: 545–551.
3 Kwok AK, Li WW, Pang CP, Lai TY, Yam GH, Chan NR *et al.* Indocyanine green staining and removal of internal limiting membrane in macular hole surgery: histology and outcome. *Am J Ophthalmol* 2001; **132**: 178–183.
4 Johnson MW, Van Newkirk MR, Meyer KA. Perifoveal vitreous detachment is the primary pathogenic event in idiopathic macular hole formation. *Arch Ophthalmol* 2001; **119**: 215–222.
5 Hisatomi T, Enaida H, Sakamoto T, Kanemaru T, Kagimoto T, Yamanaka I *et al*. Cellular migration associated with macular hole: a new method for comprehensive bird's-eye analysis of the internal limiting membrane. *Arch Ophthalmol* 2006; **124**: 1005–1011.
6 Guidry C. The role of Muller cells in fibrocontractive retinal disorders. *Prog Retin Eye Res* 2005; **24**: 75–86.
7 Tognetto D, Grandin R, Sanguinetti G, Minutola D, Di Nicola M, Di Mascio R *et al.* Internal limiting membrane removal during macular hole surgery: results of a multicenter retrospective study. *Ophthalmology* 2006; **113**: 1401–1410.

E Mendrinos[1], MK Tsilimbaris[2], P Tsoka[2] and CJ Pournaras[1]

[1]Department of Ophthalmology, Vitreo-retinal Unit, Geneva University Hospitals, Geneva, Switzerland
[2]Department of Ophthalmology, University of Crete Medical School, Heraklion, Crete, Greece
E-mail: constantin.pournaras@hcuge.ch

Eye (2010) **24,** 1121–1123; doi:10.1038/eye.2009.258; published online 23 October 2009

Sir,

Ocular complications in Mulvihill–Smith syndrome

Mulvihill–Smith is an ageing syndrome with short stature, skin nevi,[1] immunodeficiency, genital abnormalities, hearing loss, and diabetes.[2,3] Reported ophthalmological findings include astigmatism, myopia, endothelial dystrophy, keratoconus,[4] cataract,[5] amblyopia, and allergic conjunctivitis. We emphasize the unreported features in this ninth case in the literature.

Case report

A 29-year-old Japanese woman presented with decreased vision and grittiness OU. The previously reported descriptive features of this syndrome, such as progeroid appearance, microcephaly, short stature, diffuse multiple pigmented nevi on the face and trunk (Figure 1a and b), sleep disorder, sensorineural deafness, high-pitched voice, non-insulin-dependent diabetes, history of recurrent otitis media, and pulmonary infections, allowed us to diagnose the syndrome in our patient. Although immunodeficiency is a feature of this syndrome, IgG, IgA, IgM levels, PHA stimulation test, and lymphocyte subpopulation analysis were unremarkable. The new, previously unreported systemic features in the medical history of our case include severe shingles, retrolingual swelling, which proved to be a granuloma on biopsy, pseudopapillary cystic pancreatic tumour and a 2-cm right cerebellar mass (Figure 1c) found incidentally on a head MRI that remained constant in size over 3 years, and low serum Mn-SOD levels measured on two occasions (248 and 150 ng/ml, respectively; normal value: 402 ng/ml). The new unreported ocular findings include dry eye disease evidenced by elevated tear evaporation rates (6.7×10^{-7} g/cm^2/s) and low Schirmer test levels (<5 mm) OU, band keratopathy, bilateral posterior subcapsular cataracts, and meibomian gland disease evidenced by lid telangiectasia, orifice closure, and marked meibomian gland drop-out compared with normal individuals in lid transillumination

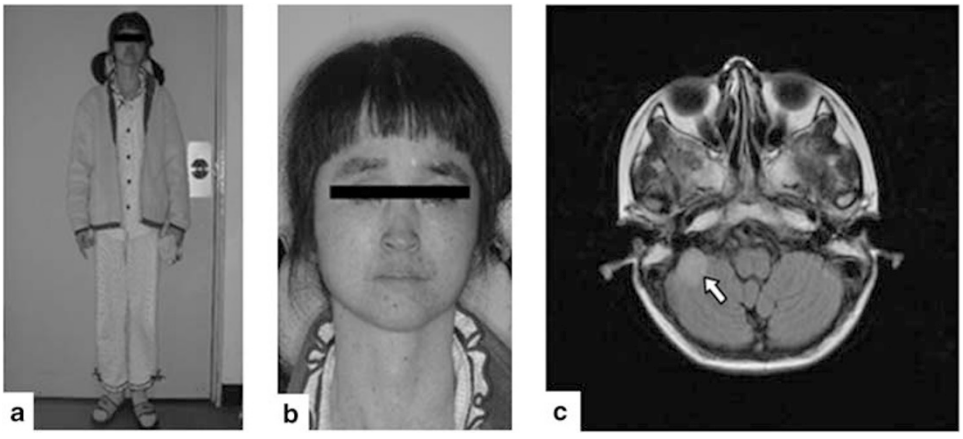

Figure 1 (a) Note the short stature. (b) Progeroid facies, multiple facial pigmented nevi, microcephaly, and mild micrognathism. (c) Magnetic resonance image (MRI) of the patient's cerebellum showing a 2 cm right cerebellar mass (white arrow).

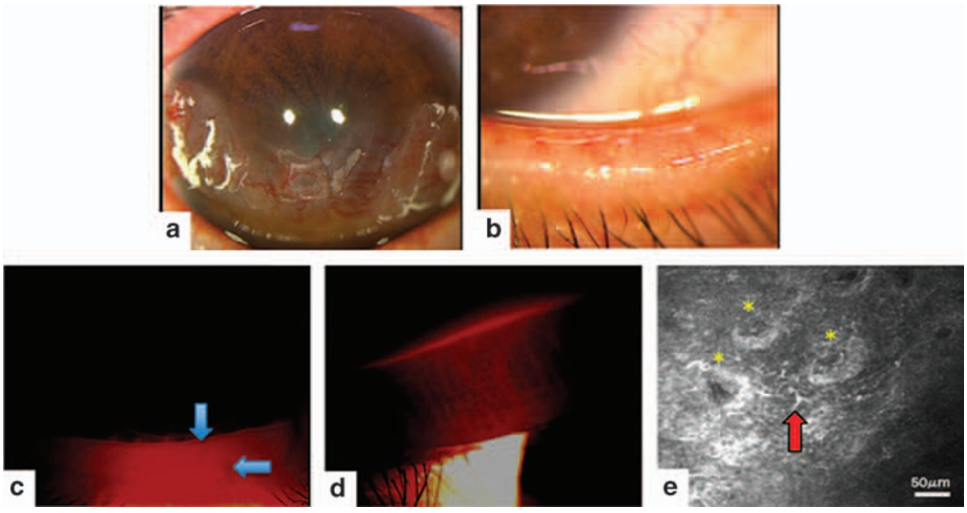

Figure 2 (a) Biomicroscopy image of the OS cornea showing band keratopathy. (b) Note the vascularization and thickening of eyelid margins with closure of meibomian gland orifices. (c) Blue arrows point to areas of meibomian gland loss in meibography. (d) Normal transillumination image of the control subject; note the intact dark rows of meibomian glands. (e) HRTII-RCM *in vivo* confocal microscopy images of the eyelids showing atrophy of the meibomian acinar units (yellow stars), with the red arrow pointing to the periglandular inflammatory cell infiltrates.

(Figure 2a–d). Confocal microscopy of the eyelids performed by us as described previously[6] also showed meibomian atrophy and inflammation (Figure 2e). The patient expired in February 2009 owing to pulmonary infection and cardiac failure.

Comment

Band keratopathy, posterior subcapsular cataract, meibomian gland, and dry eye disease are the newly described ophthalmic features of this rare syndrome, in which a sporadic mutation is suggested to be the genetic basis.[5] Oxidative stress disorder due to low Mn-SOD levels may be an explanation for the ageing manifestations of this syndrome.

Conflict of interest

The authors declare no conflict of interest.

Acknowledgements

This work was supported in part by the research grants of Johnson and Johnson Vision Care Company. The study followed the Tenets of the Declaration of Helsinki. Informed consent was obtained from the patient. This work was presented at the 33rd Japan Cornea Conference, 19–21 February 2009, Osaka, Japan. We thank Dr Junya Kuribayashi for providing the facial photographs.

References

1 Mulvihill JJ, Smith DW. Another disorder with prenatal shortness of stature and premature aging. *Birth Defects Orig Art Ser* 1975; **11**: 368–371.
2 de Silva DC, Wheatley DN, Herriot R, Brown T, Stevenson DAJ, Helms P *et al*. Mulvihill-Smith progeria-like syndrome: a further report with delineation of phenotype, immunologic deficits, and novel observation of fibroblast abnormalities. *Am J Med Genet* 1997; **69**: 56–64.
3 Ohashi H, Tsukahara M, Murano I, Fujita K, Matsuura S, Fukushima Y *et al*. Premature aging and immunodeficiency: Mulvihill-Smith syndrome? *Am J Med Genet* 1993; **45**: 597–600.
4 Bartsch O, Tympner KD, Schwinger E, Gorlin RJ. Mulvihill-Smith syndrome: case report and review. *J Med Genet* 1994; **31**: 707–711.
5 Yagihashi T, Kato M, Izumi K, Kosaki R, Yago K, Tsubota K *et al*. Case report: Adult phenotype of Mulvihill-Smith syndrome. *Am J Med Genet A* 2009; **149**: 496–500.
6 Matsumoto Y, Sato EA, Ibrahim OM, Dogru M, Tsubota K. The application of *in vivo* laser confocal microscopy to the diagnosis and evaluation of meibomian gland dysfunction. *Mol Vis* 2008; **14**: 1263–1271.

OMA Ibrahim[1,2], Y Takefumi[3], M Dogru[1,3], K Negishi[2], K Kosaki[4] and K Tsubota[2]

[1]Johnson &Johnson Ocular Surface and Visual Optics Department, Japan
[2]Department of Ophthalmology, Keio University School of Medicine, Tokyo, Japan
[3]Department of Ophthalmology, Tokyo Dental College, Chiba, Japan
[4]Department of Pediatrics, Keio University School of Medicine, Tokyo, Japan
E-mail: muratodooru@yahoo.com

Eye (2010) **24,** 1123–1124; doi:10.1038/eye.2009.280; published online 27 November 2009