Biallelic PRDM12 mutations in MiTES

This team of researchers, led by Moss, explain that midface toddler excoriations syndrome (MiTES) is a newly described condition. It is characterized by habitual scratching from the first year of life creating deep, chronic, scarring wounds around the nose and eyes. An earlier study of five siblings from a consanguineous Irish family, with lesions corresponding to MiTES plus other sensory deficits, showed homozygous mutations in a gene for hereditary sensory and autonomic neuropathy type VIII (HSAN8), PRDM12. Their aim was to study further cases of MiTES, including analysis of PRDM12. They describe five further children, from four families, with facial lesions typical of MiTES, in whom mutation analysis of PRDM12 was carried out. Homozygous or compound heterozygous pathogenic expansions of the PRDM12 polyalanine tract were found in four of five affected individuals, in three families. They concluded that the finding of autosomal recessive mutations in PRDM12 in four of five patients with MiTES extends the phenotypic spectrum of PRDM12 mutations, which usually cause HSAN8, characterized by mutilating self-inflicted wounds of the extremities, lips and tongue. By contrast, MiTES shows severe midfacial lesions with little if any evidence of generalized pain insensitivity. They conclude that this new understanding of the nature of MiTES, which can masquerade as factitious disease, will facilitate appropriate management.


Cost of narrowband ultraviolet B for psoriasis

Boswell and colleagues from Dundee explain that narrowband ultraviolet B (NB-UVB) treatment for psoriasis is considered expensive. However, existing cost data are based on estimates and do not consider indirect cost savings. Their aim in this study was to define the actual costs of NB-UVB incurred by the service provider, in addition to treatment-associated cost savings. To do this they performed data linkage of (i) comprehensive treatment records and (ii) prescribing data for all NB-UVB treatment episodes spanning 6 years in a population of 420 000. Their results are important and highly significant: National Health Service Tayside spent an average of £257 per NB-UVB treatment course (mean 257 ± 63; range 150–286 across four independent treatment sites), contrastingly sharp with the estimate of £1882 used by the U.K. National Institute for Health and Care Excellence. The cost of topical treatments averaged £128 per patient in the 12 months prior to NB-UVB, accounting for 42% of the overall drug costs incurred by these patients. This was reduced by 40% to £53 per patient over the 12-month period following NB-UVB treatment, while psoriasis-unrelated drug prescription remained unchanged, suggesting disease-specific effects of NB-UVB. The data were not the result of site-specific factors, as confirmed by highly similar results observed between treatment sites operated by distinct staff. Finally, they detail all staff hours directly and indirectly involved in treatment, allowing direct translation of cost into other healthcare systems. They concluded that NB-UVB is in fact a low-cost treatment. Furthermore, they predicted that creating or extending access to NB-UVB is likely to offer additional savings by delaying or avoiding costly third-line treatments for many patients.


Short repeated nickel exposure

Knowledge about the required duration of exposure for elicitation of allergic nickel dermatitis in nickel-allergic individuals is limited. However, it often has been proposed that short skin contact is safe. The aim of this study was to see if repeated skin contact with nickel over short time periods (3 × 10 min) could elicit allergic nickel dermatitis. Sixteen nickel-allergic adults and 10 control participants were exposed to, respectively, nickel- and aluminium-containing discs on each forearm and on each earlobe for 3 × 10 min. One arm was pretreated for 24 h with 0.5% sodium lauryl sulfate (SLS) under occlusion before exposure. Their full results are included in this issue of the BJD. This experimental study showed that relatively short repeated skin contact (3 × 10 min) with metallic nickel elicits allergic nickel dermatitis in irritated skin and at sites with previous dermatitis. The results support the restrictions in current nickel regulation.


GNAQ mutation in port-wine macrocheilia

Ma and colleagues from Shanghai explain that port-wine macrocheilia (PWM) is a congenital, progressive capillary malformation that results in soft-tissue hypertrophy in the lips. Its aetiology has not yet been fully elucidated. Their aim in this study was to investigate the frequency of GNAQ mutations in different tissues from patients with PWM, including skin, mucosa, gland and muscle, using samples obtained during cheloplasty. They carried out targeted next-generation sequencing of GNAQ on DNA extracted from 80 different affected tissues from 20 patients with PWM. They found that the GNAQ R183Q mutation was not detected in gland samples but was found in 90%, 95% and 95% of the skin, mucosal and muscle samples, respectively. The mutation frequencies in mucosa and muscle were the highest and showed no statistically significant difference (P = 0.92). They concluded that in patients with PWM, GNAQ was mutated in all tissues except for glands and that PWM is therefore congenital, and all tissue layers exhibit primary hypertrophy rather than acquired or partially related hypertrophy. They finish with an interesting and important idea: given ease of mucosal biopsy, GNAQ mutation in the lip mucosa may be a useful predictor for early-stage PWM in patients with port-wine stains affecting the lips.