human immunodeficiency virus and hepatitis status in view of the transfusions he required.

Boomslang snake antivenom is a peptin-refined immuno-globulin prepared from the serum of horses that have been hyperimmunized with boomslang (tree snake). It is commercially available.

P-50
Retrospective diagnosis of Kindler syndrome in a 37-year-old man
M. A. THOMSON, G. H. S. ASHTON,* J. A. McGRATH,* R. A. J. EADY* AND C. MOSS
Department of Dermatology, Birmingham Children’s Hospital, Birmingham and *Genetic Skin Disease Group, St John’s Institute of Dermatology, St Thomas’ Hospital, London, U.K.

Kindler syndrome is a rare autosomal recessive disorder characterized by acral blisters in infancy, periodontal disease and progressive poikiloderma. The recent finding of KIND1 mutations in Kindler syndrome facilitates early diagnosis and more precise definition of the phenotype. In the family described here, molecular diagnosis of Kindler syndrome in an infant with acral blisters clarified the diagnosis in a relative whose condition had remained obscure for 37 years. The severe adult manifestations contrast with the almost normal appearance at 12 months.

The proband, first daughter of consanguineous Asian parents, developed a few acral blisters in the neonatal period which quickly healed. She was otherwise well. The family history revealed that a cousin of her father suffered lifelong severe eczema, debilitating dental disease and a psychotic disorder. Of this cousin’s six siblings, five died before age 6 years in Pakistan, two having suffered blisters. His hospital records revealed blisters on the hands and feet from birth diagnosed as epidermolysis bullosa (EB). His poor dentition from age 3 years had been attributed to mucosal involvement with EB, and intractable constipation to the soft diet necessitated by his sore gums. In early adulthood complaints of urinary retention and dysphagia were not investigated in detail.

Examination of the proband at age 12 months revealed milia at sites of previous blisters, subtle poikiloderma on the backs of the hands and feet, healthy gums and no teeth. Examination of the cousin now aged 37 years revealed widespread reticulate poikiloderma with areas of hyper- and hypopigmentation. The dorsal aspects of the hands and feet were atrophic with a wrinkled cigarette paper-like appearance. He also had healing ulcers on the shins, nail dystrophy and severe periodontitis, but no palmoplantar keratoderma or interdigital webbing. He denied photosensitivity. Mutation analysis in both affected individuals revealed homozygosity for the common Pakistani mutation 676insC in exon 5 of KIND1 (Ashton GHS, McLean WH, South AP et al. Recurrent mutations in kindlin-1, a novel keratinocyte focal contact protein, in the autosomal recessive skin fragility and photosensitivity disorder, Kindler syndrome. J Invest Dermatol 2004; 122: 78–83).

In this family, we hope that definitive diagnosis will benefit both affected members. In the child, early diagnosis and knowledge of the severe adult phenotype should encourage preventive measures such as regular dental care and sun protection. Definitive diagnosis in the adult should facilitate future management and monitoring, and may help him at last to come to terms with his condition.

P-51
Severe hidradenitis suppurativa in a patient with a mutation of the melanocortin 4 receptor gene (MCR4)
N. KAPUR, M. SAHA, S. FAROOQI,* P. M. G. BOULOUX† AND M. H. A. RUSTIN
Departments of Dermatology and *Endocrinology, The Royal Free Hospital, London and †University Department of Medicine, Addenbrooke’s Hospital, Cambridge, U.K.

Obesity results from interactions of genetic and environmental factors. Mutations within several single genes can cause early-onset obesity, those within the melanocortin 4 receptor gene (MC4R) being responsible for the commonest monogenic cause of obesity. α-Melanocyte-stimulating hormone (α-MSH) acting on the melanocortin 4 receptor reduces food intake. α-MSH also binds to the melanocortin 5 receptor, possibly affecting sebaceous gland function. Hidradenitis suppurativa (HS) is a chronic relapsing and debilitating inflammatory disease originating in apocrine gland follicles and is commonly, although not exclusively, seen in obese individuals. We describe a severe case of HS in a patient with MC4R deficiency.

A 36-year-old Kuwaiti general practitioner presented with multiple chronic discharging areas in both axillae, neck, submammary folds, abdominal apron, inguinal and perianal regions, and buttocks. There was subcutaneous extension with induration, scarring and sinus formation. He was severely obese, weighing 230 kg, with a body mass index of 80 kg m⁻². He had been obese since the age of 5 years and had had HS for 11 years. He had type 2 diabetes mellitus and angina. Management had included numerous systemic and parenteral courses of antibiotics and surgical procedures. He had an autosomal dominant family history of obesity, with a brother dying of obesity-related diabetes and ischaemic heart disease and six other siblings weighing over 150 kg. Direct nucleotide sequence analysis of MC4R revealed our patient to be heterozygous for a missense mutation (T162I).

He was admitted and treated with oral clindamycin and rifampicin both at 300 mg twice daily, and started on a 1200-kcal diet. The lesions were treated topically with potassium permanganate soaks and then manuka honey and then flamazine. Over the course of 5 weeks he made a good recovery.

Five melanocortin receptor subtypes have been identified and the tissue expression patterns of the types are in keeping with their physiological roles. MC1R is a major pigmented control point and is mainly expressed on melanocytes; MC2R, the physiological receptor for adrenocorticotropic hormone, is expressed primarily in the adrenal cortex; MC3R and MC4R are expressed mainly in the central nervous system, the latter being important in regulation of feeding behaviour and body weight. MC5R appears to regulate sebaceous gland function.
both in animals and in humans: Hatta et al. (Hatta N, Dixon C, Ray AJ et al. Expression, candidate gene, and population studies of the melanocortin 5 receptor. J Invest Dermatol 2001; 116: 564–70) have demonstrated MC5R expression in human sebaceous, eccrine and apocrine glands. It is possible that the MC4R system may also play a role in human sebaceous gland function.

**P-52**

Papillon–Lefèvre syndrome (palmoplantar keratoderma with periodontitis): successful treatment of periodontitis, and preservation of dentition with acitretin

N.J. MORTIMER, M.J. SLADDEN AND R.A.C. GRAHAM-BROWN

Department of Dermatology, Leicester Royal Infirmary, Leicester, U.K.

A 1-year-old boy, born to consanguineous parents, presented with well-demarcated plantar erythema and hyperkeratosis on a background of low-grade eczema affecting the face and flexures. Over the next 12 months the hyperkeratosis began to involve the palms, and he developed psoriasiform patches over the extensor surfaces of the elbows, knees and knuckles. At this time he was noted to have severe chronic inflammatory periodontal disease with poor oral hygiene. A clinical diagnosis of Papillon–Lefèvre syndrome was confirmed by genetic analysis. Subsequently all the deciduous teeth were extracted and he was started on acitretin.

Papillon–Lefèvre syndrome is a rare autosomal recessive condition. Patients develop diffuse palmoplantar keratosis typically in the first 3 years of life. Many patients also have erythematous scaly patches over the elbows, knees and knuckles. The deciduous teeth erupt normally but are prematurely lost due to recurrent gingival inflammation (usually by the age of 5 years). Following eruption of secondary dentition, there is a recurrence of periodontitis leading to complete loss of teeth by the age of 16 years.

The genetic locus has been mapped to 11q14. Mutations have been found in the gene encoding a lysosomal protease called cathepsin C that lies within this region and it is thought that this may be responsible for the clinical features. Cathepsin C has an important role in the activation of certain proteases necessary for the phagocytic destruction of bacteria and local activation or deactivation of inflammatory mediators. It is also required for T cell-mediated killing.

It has been shown that systemic retinoids may be useful in the treatment of keratoderma and it has been suggested that they may be useful in the treatment of periodontitis. The mechanism of action is unknown. Our patient has been taking acitretin for 4 years with complete resolution of his keratoderma. He has a number of healthy secondary teeth, with no evidence of periodontitis. Apart from a transient rise in alanine aminotransferase, which settled with dose reduction, there have been no other complications with therapy. We hope that continued treatment with acitretin will allow the development of normal adult dentition.

**P-53**

Twins discordant for congenital cutaneous herpes simplex infection

J.E. GACH, C.J. HARVEY* AND C. MOSS

Department of Dermatology, Birmingham Children’s Hospital and *Neonatal Unit, Women’s Hospital, Birmingham, U.K.

Congenital herpes simplex virus (HSV) infection is a rare, serious and sometimes fatal disease, acquired transplacentally or from the maternal genital tract. We report cutaneous HSV in a newborn twin where the absence of cutaneous lesions in the other twin was initially thought to exclude an infective aetiology, delaying treatment.

The twin girls were born vaginally at 29 weeks to unrelated Afro-Caribbean parents. The mother had vaginal streptococcal infection during pregnancy. At birth, twin 1 had raw areas on her arms, right knee and lower left leg, reticulate silvery scars on the left upper quadrant with similar small scars scattered on the rest of the body, and prominent inguinal lymphadenopathy. After starting phototherapy for hyperbilirubinaemia on day 3, she developed clusters of 2–4 mm monomorphous vesicles on the trunk and limbs. She was otherwise well. Ophthalmoscopy revealed chorioretinitis in the left eye, brain scan was normal, and viral culture from a vesicle showed HSV type II. She was treated with intravenous aciclovir. Twin 2 had no skin lesions, but cranial ultrasound scans at 24 weeks gestation and day 1 showed gross dilatation of the lateral ventricles and a prominent massa intermedia in the third ventricle with bilateral extensive echoluent areas in the periventricular parenchyma. Eye examination showed bilateral dense symmetrical vitreous opacities obstructing the retinal view and nearly absent red reflex. These findings were consistent with HSV infection. Subsequent maternal history revealed a blistering skin rash localized to one thigh, and vaginal blistering and soreness on two occasions during pregnancy, but viral studies were not carried out. Retrospective testing of a maternal blood sample taken at booking showed anti-HSV type II antibodies.

The presence of scarring at birth suggests that fetal infection occurred in the second trimester perhaps due to maternal reinfection or reactivation of a previous HSV infection. It is not clear why maternal HSV infection led to skin and eye involvement in twin 1 and severe central nervous system but no skin involvement in twin 2.

To our knowledge, this is the first report of congenital HSV infection in twins discordant for cutaneous involvement, and is an important reminder that discordance for congenital skin lesions in twins does not exclude an infective aetiology.

**P-54**

Unrecognized scurvy caused by incorrect storage of parenteral feeds

A.TAKWALE, I.CHARONITI,* H.KELCEY,† S.PROTHEROE* AND C.MOSS

Departments of Dermatology, *Pharmacy and †Gastroenterology, Birmingham Children’s Hospital, Birmingham, U.K.

The classic dermatological signs of vitamin C deficiency are now rare, and are usually confined to individuals whose diet