

FIG. 2.—Aged 8 weeks, healing complete.

our case. Gross *et al.* (quoted by Walker *et al.*, 1960) noted the decreasing number of elastic fibres in the skin as the lesion is approached from the periphery, and suggested that during development an elastic-free skin area might be overstretched, resulting in disturbed circulation in that area and consequent maldevelopment. Duhamel (1963) has given a plausible explanation; he regards aplasia of the skin in cases such as that here described as an example of defective development of the outer layer of the body wall. Agenesis of the sternum or ribs, or of the abdominal muscles are other examples.

Most of these skin defects heal well with conservative management, and the infant does well provided there are no other congenital abnormalities.

### Summary

A case of bilateral, symmetrical thoraco-abdominal skin defect is described.

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## Hereditary pseudo vitamin D deficiency rickets in a Pakistani infant

Hereditary pseudo vitamin D deficiency rickets (Hereditäre pseudo mangelrachitis) was first described by Prader, Illig, and Heierli in 1961. The alternative name of hypocalcaemic vitamin D resistant rickets was given by them to the condition which presents during the first 2 years of life as clinical and radiological rickets with hypocalcaemia, normal or moderately reduced serum phosphate, and often mild generalized aminoaciduria. Unlike sex-linked hypophosphataemic vitamin D resistant rickets, however, there is no response to normal doses of califerol. Daily doses of 10,000 to 40,000 units (0.25–1 mg) calciferol are required to reverse the biochemical and radiological changes. Dent, Friedman, and Watson described the first British patient with the condition in 1968, and Fanconi and Prader reviewed the condition, together with a follow-up of their original cases, in 1969.

### Case report

A boy of 6 months was admitted in May 1972 for repair of an inguinal hernia. He was the second child of first generation Pakistani immigrants who are first cousins. His father came originally from the Punjab. He was a puny, miserable baby, hypotonic with only fair head control, unable to sit up without considerable support, and with marked poverty of movement. His weight was on the 3rd centile. His body was noticeably hairy with a low hairline on the forehead, and he had marked arachnodactyly, swollen epiphyses at the wrists and ankles, and a rickety rosary. He was irritable, and shortly after admission had a generalized convulsion.

At the age of 2½ months he had had three similar attacks, probably convulsions. His birthweight was just under 3 kg, he was bottle fed with a full-cream dried milk preparation (which included added vitamin D).

Radiological survey showed marked impairment of mineralization of all bones and the typical changes of active rickets. There was also a suggestion of enamel hypoplasia of the deciduous dentition.

Serum Ca 4.7 mg/100 ml, P 4.1 mg/100 ml, alkaline phosphatase 70 KA units/100 ml; Na 146 mEq/l., K 4.6, Cl 109; HCO<sub>3</sub> 20 mEq/l.; urea 34 mg/100 ml, creatinine 0.4 mg/100 ml. Serum folate 15.2 ng/ml; whole blood folate 222 ng/ml; Hb 12.4 g/100 ml. Urine calcium 4.0 mg and phosphate 280 mg in 24 hours. Urine amino acid chromatogram—moderate generalized excess of amino acids. Faecal fat 1.3 g/day over 5 days. Sweat sodium 34 mEq/l. Slit lamp examination of eyes—normal.

An initial diagnosis of nutritional rickets was made and calciferol 1000 units daily was begun in mid-May 1972. During the following week serum calcium rose to 7.5 mg/100 ml, but subsequently fell again to 4.9 mg/100 ml (Fig.). The calciferol was then increased to 10,000

September, i.e. 2½ months after starting the high dosage of calciferol.

Alkaline phosphatase took 4 months to return to normal. Serum phosphate was always normal. During his 10 weeks in hospital he gained little weight and had several pyrexial episodes for which investigations failed to find an infective cause. His hernia was eventually repaired and he was discharged home in mid-July 1972. Since then his weight has increased steadily and at the age of 1 year he looked well, and was above the 10th centile for height and weight.

His parents are first cousins, sharing a grandfather who apparently has marked bowing of the legs. There is no other history suggestive of rickets or osteomalacia in their relations in Pakistan, but their elder son, aged 2 years, has marked bowing of the legs but no radiological evidence of active rickets. His serum calcium was 10.0 mg/100 ml, phosphorus 3.1 mg/100 ml, and alkaline phosphatase 46 KA units/100 ml. Because of this he was started on 1000 units calciferol daily. His father's biochemistry is normal, but the mother has a serum calcium of 8.3 mg/100 ml, phosphorus 3.9 mg/100 ml, and alkaline phosphatase of 23 units/100 ml. Her skeletal survey is normal. The family eat about three chapatis a day, but do not give them to the two children.

### Comment

The findings in our case accord well with those reported in pseudo deficiency rickets; namely severe rickets presenting in the first year of life with symptomatic hypocalcaemia, relatively normal serum phosphate, aminoaciduria, hypotonia, and growth failure, all responding only after high doses of vitamin D with complete biochemical correction and marked catch-up growth. This contrasts with the more common hypophosphataemic sex-linked vitamin D resistant rickets that presents a little later with a similar radiological picture but with normal serum calcium and low serum phosphate, and which is not completely corrected with treatment. Even larger doses of calciferol may be required (30,000–100,000 units/day) and catch-up growth is not satisfactory. It is transmitted by sex-linked dominant inheritance. Prader *et al.* (1961) originally thought that pseudo deficiency rickets was inherited in an autosomal dominant manner. However, subsequent evidence has made an autosomal recessive inheritance more likely. The parents of our case, like several other reported cases, are first cousins, which supports this view. Another important difference is that hypophosphataemic vitamin D resistant rickets usually heals spontaneously in early adult life, whereas pseudo deficiency rickets requires life-long treatment (Fanconi and Prader, 1969; C. E. Dent, personal communication, 1972).

Rickets and osteomalacia are common in immi-

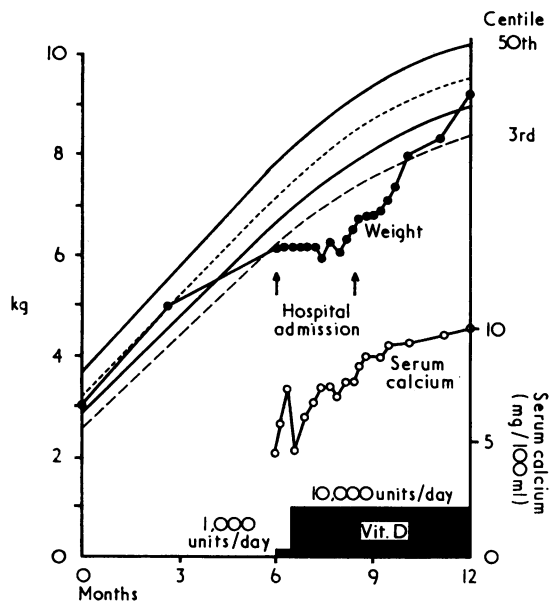


FIG.—Progress of serum calcium and weight (kg) on treatment with vitamin D.

units/day and calcium gluconate (Sandoz) 2 g/day was added to the feeds. Thereafter, the calcium rose steadily and by early July, after a month on the high dosage, had reached 7.5 mg/100 ml, though there was still no radiological sign of healing. Treatment was continued and a month later (mid-August 1972) there was no radiological sign of rickets. Serum calcium continued to rise and reached 9.6 mg/100 ml in late

grants from India and Pakistan. The reason for this is uncertain, though dietary deficiency and restricted skin synthesis of vitamin D may play a part. Wills *et al.* (1972) have suggested that the high phytic acid content in the unleavened dough used in chapatis may have aetiological significance. Swan and Cooke (1971), reviewing rickets and osteomalacia in Asian immigrants in Birmingham, found that a proportion of their patients were resistant to vitamin D, as measured by raised alkaline phosphatase after a period of treatment on conventional doses.

We speculate that this may be homozygous manifestation of a condition, which in its heterozygous form could account for the partial vitamin D resistance referred to above. The mother and brother of our case have mild biochemical abnormalities, but the father is biochemically normal.

Soriano *et al.* (1966) found normal serum levels of vitamin D-like activity in their patient when on 400 units/day. They suggested that a decreased sensitivity to vitamin D rather than malabsorption of the vitamin was the aetiological factor in their patient. Fanconi and Prader (1969) suggested that this could be accounted for by a hereditary enzyme defect resulting in metabolic block to the conversion of vitamin D<sub>3</sub> to the biologically active 25-hydroxy-cholecalciferol. However, Balsan and Garabedian (1972) discount this hypothesis, as these cases also require abnormally high doses of 25-hydroxy-cholecalciferol to achieve healing as compared with ordinary deficiency rickets. The pathogenesis, therefore, remains speculative.

The practical importance of recognizing cases of pseudo deficiency rickets is that they require lifelong high dosage of vitamin D. Severe deformity can result if treatment is not started early enough (Dent *et al.* 1968) or if it is discontinued at puberty or in early adult life (C. E. Dent, personal communication, 1972).

### Summary

A 6-month-old Pakistani boy with clinical, radiological, and biochemical features of vitamin D deficiency rickets failed to respond to calciferol 1000 units/day, and required 10 times this dosage, with added calcium, for recovery. He is considered to have hereditary pseudo vitamin D deficiency rickets which requires lifelong treatment with high doses of vitamin D. It is speculated that some cases of lesser vitamin D resistance in Asian immigrants may represent a heterozygous form of this condition; in support of this was the finding in the mother and brother of this boy of biochemical abnormalities.

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## Hydatid disease of interatrial septum

Hydatid disease due to *Echinococcus granulosus* is found in sheep farming communities throughout the world. It occurs more frequently in Wales than in other parts of the British Isles, but is a rarity even in this area. The Public Health Laboratory Service (1972) has recorded 77 cases of hydatid disease since 1966 in England and Wales, only 7 of which were in children. In 48 of the 77 recorded cases the hydatid cysts were in the liver, and in a further 17 in the lung. Only one case involving the heart was recorded. This case is presented because both the common presentation and one previously unrecorded were found in the same patient.

### Case history

The patient was a girl from a mountain sheep farm in South Wales. She presented with slight intermittent left axillary pain when 9 years old. Physical examination was normal, but a chest x-ray showed multiple rounded shadows in the left lung. The tuberculin test was negative and the Casoni test positive. An electrocardiogram and intravenous pyelogram were normal.

At thoracotomy 6 hydatid cysts were removed from the left lung and a further 10 were evacuated. 1 cyst involved the wall of the pulmonary artery but fortunately the artery wall did not rupture when the cyst was removed.