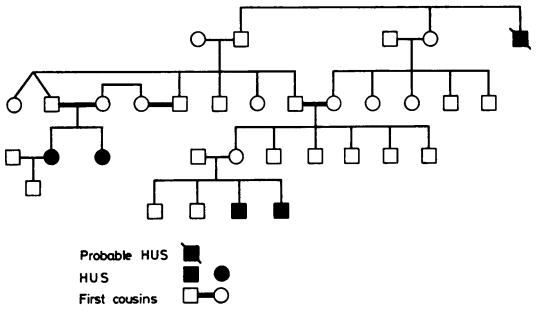

Fig. *Family tree*

the age of 24 with the diagnosis of Bright's disease. Perhaps today's diagnosis would have been thrombotic thrombocytopenic purpura or HUS. The son of Case 3 is reported to be in good health.

Discussion

The fatal outcome in this family, the study by Farr *et al.* (1975), and the reports of 3 infant sibs (Kaplan *et al.*, 1975) and 4 sibs from Zurich (Blättler *et al.*, 1975) all of whom died, together indicate a grave prognosis in cases where more than 2 family members are affected. It seems likely that the stronger the genetic evidence, the worse the prognosis. This family and its case pattern strongly support a genetic predisposition as an aetiological factor in some cases of HUS. Farr *et al.* (1975), however, were unable to identify red cell or HLA genetic markers in their family study. The inheritance pattern in this family, where first-cousin marriages were frequent, would fit an autosomal recessive rather than dominant mode of transmission. Dialysis and renal transplantation offer survival prospects to some victims, but it is doubtful whether conventional therapeutic measures for HUS can alter the outcome in these cases.

Summary

Two pairs of cases of HUS are reported from two generations in a family where first-cousin marriages have been frequent. All 4 died. We suggest that there is a high expected mortality in those familial cases of HUS where genetic factors are strongest. An autosomal recessive inheritance pattern is suggested in this family.

We thank Dr F. S. W. Brimblecombe for permission to report the 2 younger cases, and for help with this paper.

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Congenital heart block and hypothyroidism

Complete heart block has been reported in association with thyrotoxicosis (Stern *et al.*, 1970; Kernoff *et al.*, 1973; Fairfax and Leatham, 1975), and hypothyroidism (Singh *et al.*, 1973; Fairfax and Leatham, 1975) in adults. Autoimmunity (Zoob and Smith, 1963) and myocarditis (Hudson, 1965) have been implicated. I describe a case here in which hypothyroidism was associated with congenital heart block. I have not been able to discover another such case in the literature.

Case report

The mother of the patient, aged 30, blood group B Rh positive, had a previous normal child. She developed rheumatoid arthritis after his birth and was treated with aspirin alone. She became pregnant again soon after diagnosis. There is no relevant family history.

In this pregnancy polyhydramnios was noted by the obstetrician at 36 weeks and she had mild oedema of the legs at 38 weeks' gestation, treated with hydrochlorothiazide, reserpine, and potassium chloride

(Salupres), one tablet twice a day. Delivery was induced electively at term by forewater amniotomy and oxytocin drip. Fetal heart was noted to be steady at 130–140/minute. With the change of nursing staff in the labour ward a 'drop' in fetal heart was noted from 132/min to about 68/min, and then to about 30–40/min. Midcavity forceps were applied and a girl was delivered quickly because of 'fetal distress'. Mother had pethidine 100 mg and promazine 25 mg 6½ hours before delivery.

Baby's condition was good at birth. Apgar score was 8 at one minute and 9 at five minutes. Heart rate was 60/min with extra beats. A midsystolic murmur was heard along the left lower sternal edge and at the apex. Birthweight was 3·365 kg, length 50 cm, head circumference 36 cm. She went home after 8 days weighing 3·38 kg on breast feeding. Electrocardiogram (Fig. 1) showed complete heart block. Chest *x*-ray showed a large heart with well aerated lung fields. Mild jaundice was noted on the third day but it cleared without treatment by 6 to 7 days. The probable explanation of the 'drop' in fetal heart rate was that nurses were counting two heart sounds as two heart beats because of slow heart rate. When the heart rate was correctly counted it was thought to have 'dropped' from a steady 130–140/min to 68/min, i.e. to exactly half.

The baby was followed up for congenital heart block. At 1 month she looked well, was feeding well on the breast, and was gaining weight. Heart rate was 40/min and weight 4 kg.

At 3 months she was seen again for a history of poor feeding, sleeping most of the time, not having smiled, and not taking much notice of the surroundings. She was not constipated. On examination she looked hypothyroid (Fig. 2*a*). Her tongue was

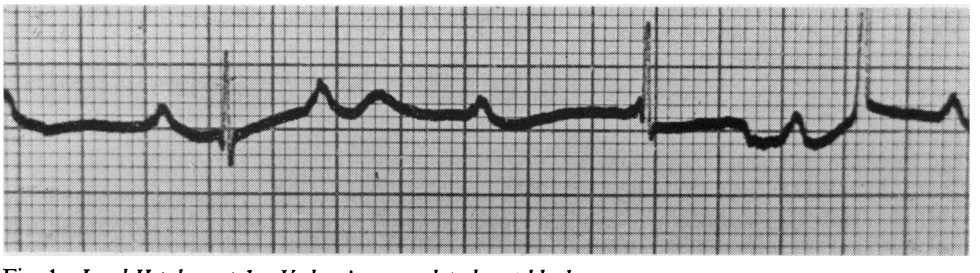

Fig. 1 *Lead II taken at 1 mV showing complete heart block.*

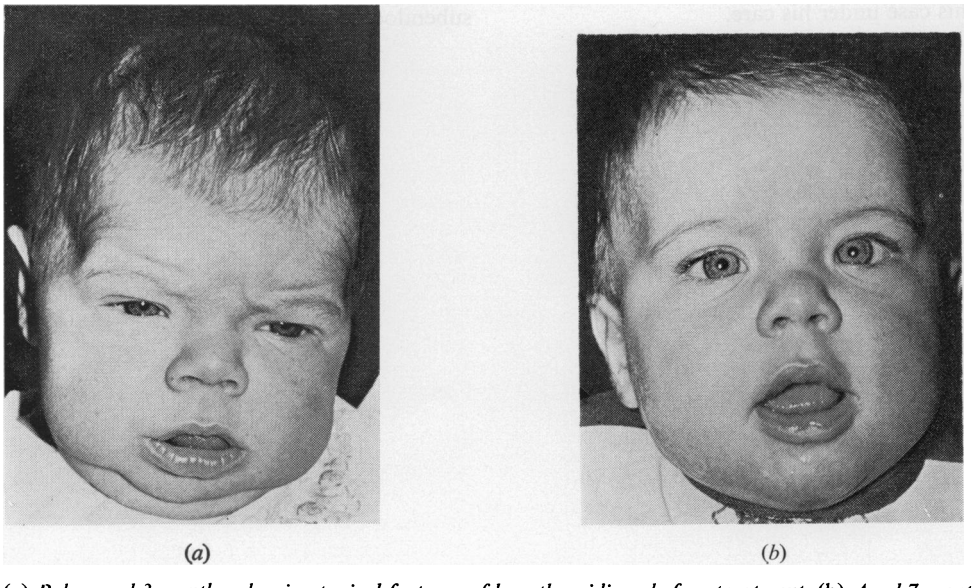

Fig. 2 (a) *Baby aged 3 months, showing typical features of hypothyroidism, before treatment.* (b) *Aged 7 months, 4 months after starting treatment, showing squint but otherwise normal features.*

large, she had a hoarse cry, her hair was not coarse, her skin was smooth. Heart rate was 48/min. Thyroid function tests done by competitive protein binding and *x*-ray for bone age confirmed the diagnosis. Serum thyroxine $<0{\cdot}5$ μg/100 ml ($<6{\cdot}4$ μmol/l); free thyroxine index $<0{\cdot}5$ μg/100 ml ($<6{\cdot}4$ μmol/l); triiodothyronine uptake 147; thyroid-stimulating hormone, >30 ng/l; cholesterol 275 mg/100 ml (7·5 mmol/l). No ossification of upper femoral epiphysis or at the wrist, suggesting delayed maturation.

She was started on rapidly increasing doses of thyroxine starting from 0·025 mg daily, going up to 0·175 mg daily in 2½ months. Heart rate remained between 48 and 60/min.

At one year she was sitting unaided, rolling over, reaching out for toys, said 'dad-dad', 'mum-mum' etc., had 7 teeth, and was bright and alert (Fig. 2*b*). Development was consistent with the age of 9 months. Heart rate was 48/min. Electrocardiogram has shown no change. She has a squint which is being treated by an ophthalmologist.

Summary

A case is described in which congenital heart block was associated with hypothyroidism. The hypothyroidism is being treated successfully with thyroxine, but this has had no effect on the heart block.

I am grateful to Dr E. H. Back for allowing me to report this case under his care.

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Note added in proof: The baby died suddenly on 8 November 1977. She was seen on 4 November and was extremely well, walking and talking, her development nearly normal. She still had a complete heart block and presumably had sudden arrhythmia which resulted in her death. Necropsy showed evidence of subendocardial fibroelastosis.