which can be excluded on the basis of additional skin or limb abnormalities, such as in Roberts's pseudothyaldomide syndrome or incontinentia pigmenti. The normal karyotype in our cases excluded chromosomal abnormalities and there was no evidence of a congenital rubella embryopathy.

When hypogonadism occurs in addition to microcephaly, mental retardation, and cataracts, only a small number of syndromes needs to be considered in the differential diagnosis. One is Smith-Lemli-Opitz syndrome, but cataracts in that syndrome are rare and, looking at table 2, there are more differences than similarities. The other condition that needs excluding is the oculopatrotocerebral syndrome, in which a cleft palate and a persistent hyperplastic primary vitreous with microphthalmia are cardinal features, but which are absent in Martsolf's syndrome.

Lowes's syndrome, which is X linked, was excluded by finding a normal amino acid pattern and galactosaemia was also biochemically excluded.

A number of other minor features were originally reported by both Martsolf et al and Sanchez et al. These include brachycephaly, lax finger joints, talipes valgus, a pouting mouth, and maxillary retrusum with slight hirsutism, but these are difficult to evaluate. The facial features in the two sibs in this report are not distinct. Both are clearly microcephalic and have small jaws with slight hirsutism, but it remains uncertain whether facial dysmorphic features are useful in helping the clinician to establish the diagnosis.

It is of interest that one of the sibs had a cardiomyopathy and the other died from cardiac failure, although the exact nature of the lesion was not elucidated. The combination of cataracts, a cardiomyopathy, and nephronophthisis has been described in mitochondrial cytopathy. The child described in that report had typical features of a mitochondrial myopathy on muscle biopsy, whereas case 1 in our family had a muscle biopsy which was normal. Microcephaly in the neonatal period would also be very unusual in mitochondrial disease.

In summary, we report a brother and sister pair with features of Martsolf's syndrome to indicate its recessive nature and also to suggest that, in view of the presence of a cardiomyopathy in one sib and cardiac failure in the other sib, the spectrum is either larger than first anticipated or alternatively that the condition described in this paper is a separate genetically determined entity.

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Dominant inheritance of Scheuermann's juvenile kyphosis

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SUMMARY We report a family in which Scheuermann's juvenile kyphosis is present in three successive generations with male to male transmission. This provides further evidence for autosomal dominant inheritance in at least some families with this condition.

Scheuermann's disease (MIM18144), or spinal osteochondrosis, although probably first recognised by Stafford in 1832, was described by Scheuermann in 1920. It is a condition where there is an abnormal and fixed thoracic kyphosis of unknown aetiology that develops around the time of puberty. Associated with this physical deformity, Scheuermann found radiographical evidence of thoracic vertebral wedging and irregularities of the growth plates of the affected
Case reports

A thoracic kyphosis is commonest with the summit usually in the area T6 to T11 and Scheuermann also noted a coexistent mild scoliosis in 75% of cases.

This is the most frequent cause of adolescent kyphosis with a population incidence of 0.4% to 8.3% depending upon whether the diagnosis was based, respectively, upon clinical or radiographical criteria, since not all are symptomatic.

Case report

A 14 year old boy (GLA7773) was referred for orthopaedic assessment as he had developed a round back deformity. The deformity was first noticed at the age of 13 and had been progressive. His father had been particularly concerned as both he and his own father had been similarly affected.

On examination, the boy was 170 cm tall, with a span of 170 cm and, in addition to a round back deformity, had a mild scoliosis to the left (fig 1). The deformity appeared relatively fixed with an exaggerated lumbar lordosis.

Radiographs confirmed that he had spinal

FIG 1 Photograph of index case and his father.

FIG 2 Lateral spine radiograph of index case.

FIG 3 Lateral spine radiograph of father.
osteochondrosis as there was irregularity of the growth plates and wedging of the mid-thoracic vertebrae producing a fixed kyphosis of 58°, which only corrected to 49° on extension (fig 2). He had a scoliosis from T8 to L1 to the left measuring 10°. Correction of the kyphosis was attempted using a Milwaukee brace.

In the family history his father had developed a round back deformity which had been static for 15 years despite no active treatment, and examination showed a similar but more severe deformity (fig 1). A radiograph of his spine confirmed that he also had Scheuermann’s disease (fig 3).

The paternal grandfather had developed a similar round back deformity in his teens and a diagnosis of spinal osteochondrosis was confirmed both clinically and radiologically (figs 4 and 5).

Discussion

Scheuermann’s disease clearly shows familial clustering. Of seven sets of monozygotic twins collected by Scheuermann,² six were clinically concordant, and Bjersand³ also described a concordant monozygotic twin pair.

Sorensen⁴ studied 151 sibs belonging to 79 of the 103 families he had collected and found that more than 48% of all sibs had Scheuermann’s disease. It was also found in 65% of parents with a slight preponderance of fathers, and in 86% of cases the site of the kyphosis was concordant.

Halal et al⁵ described five families in which the disease seems to follow an autosomal dominant pattern of inheritance. The diagnostic criteria described by Bradford et al⁶ were present in at least one affected member of each family.

Arpad et al⁷ examined the inheritance, using clinical and radiological means, of 78 index cases during epidemiological screening and of 198 first degree relatives and found that the disease followed an autosomal dominant pattern of inheritance with nearly complete penetrance.

In one remarkable family, Kewelramani⁸ presented clinical and laboratory findings from a family where Scheuermann’s kyphoscoliosis and Charcot-Marie-Tooth syndrome cosegregated at five of six meiotic events. Nielsen and Pilgaard⁹ also described a possible variant where spinal osteochondrosis and hereditary juvenile anterior fusion of the vertebral bodies in the thoracolumbar region coexisted and were inherited in a pattern consistent with autosomal dominant inheritance.

The family reported here and other published cases would support autosomal dominant inheritance in a substantial proportion of cases of Scheuermann’s disease.
Case reports

mamm’s disease. Since in severe cases early diagnosis and treatment lead to a more favourable outcome, it is therefore important that the offspring of an affected person be examined carefully around puberty.

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Prenatal diagnosis of the megacystis-microcolon-intestinal hypoperistalsis syndrome

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SUMMARY The ultrasonographic and necropsy findings in a male fetus with the megacystis-microcolon-intestinal hypoperistalsis syndrome are reported. The presence of vacuolation and degeneration in smooth muscle of bowel and bladder wall supports a previous suggestion that the macroscopic findings in this syndrome are the consequence of an underlying visceral myopathy. The unusual degree of severity of the findings in this fetus may explain the marked skewing of the sex ratio observed in affected liveborn infants.

The megacystis-microcolon-intestinal hypoperistalsis (MMIH) syndrome is a rare condition in which the underlying pathogenesis is poorly understood. Affected sibs have been reported on five occasions1–5 and there have been two reports of parental consanguinity,6 6 so that autosomal recessive inheritance is highly probable. However, a review of published reports indicates a striking excess of affected females over males. We now describe the prenatal and necropsy findings in an affected male fetus which offer some insight into the likely explanation for this apparent skewing of the sex ratio.

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Case report

The fetus was the product of the third pregnancy of healthy, unrelated parents. Their first child was a healthy female. The next pregnancy resulted in the delivery of a male infant who died at the age of four hours as a consequence of multiple abnormalities, which were believed to include urethral atresia and possible intestinal atresia. Necropsy was not undertaken.

Because of the absence of a clear diagnosis in this baby, the parents requested that their third pregnancy be monitored by ultrasonography. At 18 weeks a section through the fetal abdomen showed a markedly distended bladder with ascites and oligohydramnios (fig 1). Termination of pregnancy was requested. The male fetus was noted to have a grossly distended abdomen with no other obvious external anomalies. At necropsy the small and large bowel were malrotated with the caecum and appendix in the right hypochondrium. The colon was very narrow in diameter (fig 2) with a patent lumen. Small strands of mesentery were wound around the caecum, appendix, and duodenum. The liver and gallbladder were normal and the presence of gross ascites was confirmed. Both kidneys were small and dysplastic. The ureters were mildly dilated at their distal ends and the bladder was grossly distended (fig 3). Normal male genitalia were present.
Dominant inheritance of Scheuermann's juvenile kyphosis.
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