Case reports

mann'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.
FIG 1 Longitudinal ultrasonographic section through the fetal abdomen at 18 weeks. The arrows from left to right indicate distended bladder, ascites, and intestine.

FIG 2 Postmortem view of the small and large intestine showing the microcolon.

FIG 3 Transverse section through the bladder showing gross thickening.
Histology of the colon showed the outer muscle coat to be very thin with vacuolar muscle degeneration and normal ganglia. In the bladder wall there was abundant loose connective tissue separating groups of muscle fibres (fig 4), which showed vacuolation and degeneration similar to that seen in the microcolon. The urethra was fully patent.

No other internal anomalies were found. The only abnormality shown by skeletal survey was mild flaring of the lower ribs secondary to the abdominal distension. Chromosome analysis of cultured lymphocytes showed a normal male karyotype.

Discussion

This is believed to be the first reported case of prenatal diagnosis of the MMIH syndrome resulting in termination of pregnancy. Although the precise diagnosis was not apparent at the time, subsequent necropsy studies strongly suggest that the fetus is likely to have had this condition. This conviction is based on (1) the absence of any urethral obstruction on careful dissection, (2) the presence of bladder hypertrophy in association with a microcolon, and (3) histological evidence of vacuolar degenerative changes in smooth muscle of colon and bladder, as documented in two newborn infants by Puri et al.\(^7\) Malrotation\(^1,2,4\) and unusual mesenteric adhesions\(^8\) have also been noted previously.

Prenatal ultrasound findings in the MMIH syndrome have been described before in the second trimester,\(^5,9\) but in these cases the absence of oligohydranmos prompted hope of a reasonable prognosis, although in this condition the ultimate long term prospects for survival are extremely poor. Thus, this case is unusual, not only because of associated oligohydranmos, but also because of ascites and renal dysplasia, which are not thought to be characteristic of the MMIH syndrome. However, any or all of these features may be found in babies with the ‘prune belly’ syndrome, a malformation complex usually attributed to the consequences of urethral obstruction.\(^10\) This raises the possibility that the disease process in the MMIH syndrome may sometimes be much more severe in males resulting either in spontaneous mid-pregnancy loss, or, as suggested by Winter and Knowles,\(^4\) in misdiagnosis as examples of the aetiologically heterogeneous ‘prune belly’ syndrome, a condition or group of conditions known to be much more common in males. Either of these possibilities would adequately explain the distortion of the sex ratio observed in the MMIH syndrome.

Note added in proof

Penman and Lilford\(^11\) have recently provided details of another case of the MMIH syndrome diagnosed prenatally.

References

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