gressive in the two children followed up for some time, one for 8 years and one for 22 months. Two patients, KB and ME, showed marked developmental delay which we feel goes beyond the effect of β -glucuronidase deficiency.^{2,4} Both of these patients had neonatal hyperbilirubinaemia. KB developed a giant cell hepatitis and a downhill clinical course with death at 2 years 9 months and ME developed neurological signs of kernicterus.

Onset of corneal opacification varied from 7 months³ to 8 years.⁴

Progressive joint contractures, first manifest in the newborn period, have been seen in one patient, the subject of this report.

Findings of dysostosis multiplex have been variable. Consistent findings have included a 'J-shaped' sella and characteristic pelvic abnormalities with acetabular dysplasia, narrow sciatic notches, and hypoplastic basilar portions of the ilia.¹⁻⁴ Widening of the ribs has been noted in three patients,^{1,3} and pointed proximal metacarpals have been described in two.¹ Vertebral abnormalities have differed: a hypoplastic odontoid and shortening and anterior irregularities of the vertebral bodies occurred in one child^{1,4}; wedge deformities of the lumbar vertebrae were described in another.³ Anterior inferior beaking of the lower thoracic and lumbar vertebrae was noted in the subject of this report. In one additional patient the spine was reportedly normal, although a mild gibbus deformity was apparent.² Hip dysplasia has been noted in two patients^{1,4} and has been severe and progressive in one child.⁴ Other abnormalities have included medullary expansion of the proximal humeri¹ and abnormal irregular ossification of the humeral heads.

Frequent respiratory infections have been described in all patients.¹⁻³

The variability in urinary acid mucopolysaccharide excretion exhibited by this child merits comment. The two screens were carried out in the same laboratory using the same procedure. Although we cannot rule out a problem with faulty preservation of the first urine sample, variable urinary acid mucopolysaccharide excretion in other patients at various points in time has previously been encountered (Sly, 1980, personal communication).

In summary, this patient's course and data from published reports indicate that MPS VII, unlike the other known mucopolysaccharidoses, is a distinct clinical entity recognisable in the newborn period. Hydrocephalus and progressive joint contractures are occasional features. With respect to developmental prognosis, this disorder is most likely to be associated with moderate mental deficiency which does not progress over time.⁴

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Anal atresia and the Klein-Waardenburg syndrome

SUMMARY A 3-month-old male infant with type I Klein-Waardenburg syndrome with an imperforated anus and a perineal fistula is reported. The possible association of this gastrointestinal malformation with the KW syndrome is discussed.

The most common form of the Klein-Waardenburg (KW) syndrome is characterised by lateral displacement of the medial canthi and lacrimal punctae.¹ The

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KW syndrome type I is inherited as an autosomal dominant disorder with varying penetrance and expressivity.^{2,3} The purpose of this brief report is to call attention to a previously undescribed anomaly, anal atresia, which may be associated with this syndrome.

Case report

A 3-month-old male infant was referred to us because of a 2-week history of constipation accompanied by mild rectal bleeding. The parents were

FIG 1 Family pedigree.

consanguineous Oriental Jews, originating from Iraq (fig 1). The child was born at term after a normal pregnancy and delivery. Apgar score at one minute was 9. Birthweight was 3620 g. There were no perinatal or neonatal difficulties and the baby was discharged from the nursery in good condition.

Upon physical examination, the child appeared to be an alert infant with a weight of 6100 g (75th centile), a length of 63 cm (90th centile), and a head circumference of 41 cm (75th centile).

The main physical findings included lateral displacement of the medial canthi and lacrimal punctae, broad nasal root, hypoplastic nasal alae, right preauricular sinus, cupid-bow configuration of the upper lip (fig 2a, b), areas of vitiligo and hyperpigmentation of the skin about the abdomen, perineum, and left leg, and clinodactyly of the fifth finger of the left hand. A white forelock was absent. Examination of the perineum revealed an imperforate anus with an anal dimple and a perineal fistula situated 1 cm anteriorly (fig 2c). Chromosomal analysis showed a normal male karyotype. Intravenous pyelography showed no urological anomalies. Barium enema confirmed the presence of perineal fistula and rectal atresia and showed agenesis of the fifth sacral vertebra and the coccyx (fig 2d). The infant was treated with mineral oil and passed stools daily without difficulty. He was then referred to the Department of Pediatric Surgery. An audiogram done at 4 months of age showed no definite hearing impairment and it is planned to repeat this examination when the child is older.

FIG 2 (a) (b) The proband. Note the lateral displacement of the medial canthi, broad nasal root, hypoplastic alae, and cupid-bow configuration of the upper lip; (c) imperforate anus with an anal dimple and a perineal fistula; (d) radiograph showing rectal atresia and agenesis of the fifth sacral vertebra and coccyx.

Discussion

The KW syndrome was diagnosed in our patient on the basis of presence of such classic features as lateral displacement of the medial canthi and lacrimal punctae, broad nasal root, hypoplastic nasal alae, cupid-bow configuration of the upper lip, and areas of skin hyperpigmentation and vitiligo.

Despite the fact that our proband's parents were consanguineous, we believe that he has type I KW syndrome which is transmitted as an autosomal dominant disorder and as such he represents a new mutation.

The most frequently observed gastrointestinal malformation known to be associated with the KW syndrome is aganglionic megacolon (Hirschsprung disease).⁴ The clinical association between the KW syndrome and aganglionic megacolon fits well with the general hypothesis that this syndrome results from a defect in migration of the neural crest cells.⁴ This case report represents the first description of a patient with the KW syndrome type I with anal atresia. His other malformations, consisting of agenesis of the fifth sacral vertebra and coccyx, are frequently observed in patients with various types of anorectal malformation.⁵ In 1978, Pinsky⁵ reviewed the syndromology of the various anorectal malformations (atresia, stenosis, ectopia) and no mention was made of an association of any of these with the KW syndrome. However, it should be noted that these anal malformations are relatively rare (1:10 000)⁶ and perhaps such an association has been overlooked.

Fisch,⁷ in 1959, reported on one patient with the KW syndrome who had oesophageal atresia.

At present it is not possible to state that either anal or oesophageal atresia is aetiologically linked with the KW syndrome as is aganglionic megacolon.⁴ Future clinical observations regarding atretic lesions of the gastrointestinal tract and the KW syndrome will aid in clarifying this question.

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