

Central nervous system malformations in Mohr's syndrome

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SUMMARY A boy with severe developmental delay, bilateral, symmetrical hallucal duplication, and accessory alveolar frenula was found to have radiological evidence of a large arachnoid cyst compressing the cerebellum and brain stem. We review neurological abnormalities in Mohr's syndrome.

The oro-facial-digital syndrome (OFD) type II, or Mohr's syndrome, has been designated, as a separate entity from OFD type I, on the basis of autosomal recessive inheritance, bilateral hallucal reduplication, normal hair and skin, and the occasional presence of a conductive hearing loss.¹⁻³ There has been a small number of reports of neurological features in association with Mohr's syndrome, but in general patients with OFD type II differ from type I in that intelligence is usually normal and central nervous system malformations are unusual.^{4,5} We report a

case with features of Mohr's syndrome in a patient who had a large posterior fossa subarachnoid cyst and occipital meningocele, which have not previously been described in this condition.

Case report and investigations

The proband, a boy, was the fourth child born to healthy, non-consanguineous, Irish parents. The pregnancy was remarkable only in that the mother noticed reduced fetal movements throughout. Spontaneous vaginal delivery took place at 38 weeks. Birth weight was 3400 g. He was noted to be hypotonic and clinical suspicion of a meningocele was confirmed radiologically. He was a poor feeder and early in life developed marked apnoeic episodes. These were particularly prone to occur when hungry or otherwise aggravated and were often associated with cyanosis. Marked hypotonia followed these episodes but no seizures were noted. The meningocele was excised at the age of three months.

His developmental milestones were markedly



FIG 1 Accessory alveolar frenula.

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Lingjun Ying

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