#### Case reports

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## 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. <sup>1-3</sup> 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. <sup>4-3</sup> 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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**BM King** 

Recent Advances in the developmental biology of central nervous system malformations ,1979 The Developing Brain and Its Disorders M. Arima, Y. Suzuki, H. Yabuuchi, 1985-04-09 **Current Catalog** National Library of Medicine (U.S.),1979 First multi year cumulation covers six years 1965 70 **DHHS Publication** ,1980 Reproductive and Perinatal Epidemiology Michele Kiely, 2019-06-04 First Published in 1990 Reproductive and Perinatal Epidemiology reviews topics that are central to reproductive and perinatal epidemiology as well as the methodological issues surrounding research in maternal and child health Important topics covered include prenatal care Sudden Infant Death Syndrome SIDS epidemiology of neonatal brain hemorrhage early pregnancy methodological issues in drug epidemiology environmental exposures and reproduction and occupational exposures and reproduction This book is essential reading for clinicians researchers obstetricians pediatricians and graduate students in public health **Published Scientific Papers of the** National Institutes of Health National Institutes of Health (U.S.), 1981 Each issue lists papers published during the preceding year **Child Health and Human Development** Tom James Truss, 1981 **Cumulated Index Medicus** .1969 Human Genetics, Informational and Educational Materials National Clearinghouse for Human Genetic Diseases (U.S.), **Toxicology Research Projects Directory**, 1980-10 **Human Genetics** .1980 Sex Differentiation and Chromosomal Abnormalities, 1978 Bibliographic Guide to Conference Publications New York Public Library. Research Libraries, 1979 Vols for 1975 include publications cataloged by the Research Libraries of the New York Public Library with additional entries from the Library of Congress MARC tapes

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