Hypoglossia-hypodactyly syndrome with hydrocephalus

We note with interest the report by Gillioret et al. suggesting that a case of hypoglossia-hypodactyly syndrome with hydrocephalus was caused by an acquired inflammatory process. The authors indicate that their case was an example of Hanhart’s syndrome.

The histopathology in the brain stem of their case is very similar and probably identical to that reported in two cases of Moebius syndrome reported by ourselves and by a variety of other authors documented in that paper. Although two cases in their series were associated with high fever, one with pneumoni-
influenza and the other with an abscessed wisdom tooth, an observation confirmed by Graham et al. at a similar gestational stage, there is no evidence that these infections involved the fetus or membranes, and ample evidence of a uterine origin. An acquired and persistent association with waves added to the already well-known history of meconium aspiration, prolonged rupture of the membranes, previous uterine surgery, and alcohol abuse as well as hyperthermia. A rat model in which the uterine artery is occluded or handled and the uterus itself handled, in addition to systemic administration of cocain, results in similar brain stem lesions. Other animal experiments include the work of Miller and Myers, who simultaneously occluded the major vessels of adult rhesus monkeys for a measured length of time. During arrest of systemic flow, the pulmonary and coronary vessels remained unobstructed allowing the passage of oxygenating blood through the lungs and heart. The animals, who did not have significant post arrest hypotension, had a typical pattern of brain stem ischemia and necrosis. Ranck and Windle2 caused asphyxia in near term Macaca mulatta by detaching the placenta at hysterotomy and keeping the fetal mem-


During pregnancy could be associated with probatory drug intake. The drugs associated with these fragility X cases included the thalas-
sis and the drug, and Bectenec, which have shown to be non-teratogenic. The con-
sanguinity noted by them in previous reports is also probably coincidental, although con-
genital semi-thalassemia and neural petal hypoplasia associated with limb deficiency have been well recorded in some cases to be monogenic in nature, either autosomal dominant or X linked recessive.

We would also wish to draw attention to previous correspondence on this issue in this Journal in 1990.1

A H LIPSON

Genetics and Dysmorphology Unit, The Children’s Hospital, Pyrmont Bridge Road, Campedown, Sydney 2050, Australia.

D D WEAVER

Indiana University School of Medicine, Indianapolis, Indiana, USA.

1 Gillioret Y, Van Maldegem L, Chef R, Koulouchi L. Hypoglossia-hypodactyly syn-

2 Lipson AH, Webster WS, Brown-Woodman PDC, Osborne RA. Moebius syndrome: an-

3 Graham JM, Edwards MJ, Lipson AH, Webster WS, Edwards MJ. Gestational hyperthermia as a cause for Moebius syn-


7 Ranck JB, Windle WF. Brain damage in the monkey, macaca mulatta, by asphyxia neon-

8 Dambrosia ML, Drygik T, Szretter J, Woznie-
wicz J, Meyers RE. Topography of lesions of brain stem ischemia and necrosis. Ranck and Windle2 caus-
author:2023-09-14 by guest. Protected by copyright. Edition in this laboratory. For some years there has been a marked decrease in the number of fragile X cases reported in the literature.2 We have concluded that the incidence in the general population is not as high as the figure usually quoted (that is, in the region of 1 in 1000).

The laboratory began routine screening for fragile X during 1980 and in the 10 year period from 1980 to 1990 a total of 47 positive cases was identified. Forty-three of these were from families with a history of X linked mental retardation, including 13 cases from one large kindred.2 Only four patients with no family history were positive.

In the first three years of the study most of the referrals were from the families with a positive history. However, after the initial interest waned, and recall of patients seen before the discovery of the fragile X chromo-

33.5% of the total referrals. Only 2.2% of the single case referrals were positive compared with 32.8% of the familial cases. The number of positive cases peaked in 1983 and have fallen off since then. Only one case was detected in 1988 and one in 1989. There were no positive cases in 1990.

The number of births in the Cape Town hospital area is approximately 2500 children. In the 10 year period studied, 2500 children were in the age group for referral. If the incidence is taken as 1 in 1000 we would have expected 250 cases of fragile X over this period. Only 47 cases were detected.

The question asked by Webb and Bundey was whether the incidence of fragile X had fallen off, or was more education of hospital staff required. In our situation the majority of referrals are from hospital clinics (neuro-

do not care whether the incidence of fragile X had fallen off, or was more education of hospital staff required. In our situation the majority of referrals are from hospital clinics (neuro-


