Wildervanck’s syndrome with bilateral subluxation of lens and facial paralysis

SUMMARY A 15-year-old female was found to have the typical features of Wildervanck’s syndrome, including Klippel-Feil anomaly, abducens paralysis, retraction of the bulbi, and deafness. In addition, she had bilateral lens subluxation and facial paralysis, neither of which have been reported in patients with Wildervanck’s syndrome.

Case report

The proband was a 15-year-old female, born to a 29-year-old G4, P3, A1 mother, following an uncomplicated term pregnancy with no exposure to agents known to be teratogenic. Delivery was uncomplicated and spontaneous at term. Her birth weight was 3.5 kg. In the first month, the parents noted facial asymmetry. There were no relevant problems until the age of 6 years, when a diagnosis of facial paralysis and fusion of cervical vertebrae was made. The parents were healthy and consanguinity was denied; a younger sister and brother of the patient were normal. There was no family history of any congenital malformation, deafness, or neurological abnormality. Physical examination revealed a well developed girl. She weighed 76 kg (90th to 95th centile), was 156 cm tall (10th to 25th centile), and occipitofrontal circumference was 55 cm (50th centile). She had facial asymmetry (fig 1), shortness of the neck, pterygium colli, and a low posterior hairline. Neurological examination showed bilateral abducens and right facial paralysis, but other neurological alterations, such as hemiplegia, quadriplegia, or paralysis of other cranial nerves, were absent. Ocular examination showed bilateral temporal subluxation of the lens (fig 2) and retraction of the bulbi. Pupillary size, shape, and reactions were normal, as were the eyelids, corneae, and fundi. An x-ray examination showed cervical scoliosis and fusion of the second, third, and fourth cervical vertebrae (fig 3). In addition, the patient had bilateral sensorineural deafness. Routine laboratory studies showed normal urine analysis and blood count and blood amino-acid analysis was also normal.

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Case reports

Discussion

A syndrome of Klippel-Feil anomaly, deafness, abducens paralysis, and retraction of the bulbi has been described by Wildervanck and Wildervanck et al. Although the syndrome was thought to be responsible for at least 1% of deafness among girls, there have been only a few reports of this syndrome in recent years. Moreover, to our knowledge, there are no reports of the association of bilateral subluxation of the lens and unilateral facial paralysis in association with this syndrome. It is possible that the facial paralysis resulted from birth trauma although there was no evidence for this in the history. It is even more difficult to understand lens subluxation in this patient, since we have excluded most metabolic diseases associated with subluxation of the lens, including homocystinuria, sulphite oxidase deficiency, and hyperlysinaemia.

To our knowledge there are no other published reports of developmental syndromes with the clinical features found in our patient. Therefore, either the findings of lens subluxation and facial paralysis are just a coincidence, or they represent previously undescribed associated defects in this rare syndrome. Other reports of this syndrome include other infrequently seen signs, such as pseudopapilloedema and occipital meningcele, but not subluxation of the lens and facial paralysis. The syndrome is genetically limited to females which suggests X linked dominant transmission with lethality in the hemizygous male. Other authors think that multifactorial inheritance is more likely. Further reports are necessary to establish the incidence of subluxation of the lens and facial paralysis in this syndrome.

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δβ-thalassaemia in Sicily: report of a case of double heterozygosity for Aγδβ-thalassaemia and AγGγδβ-thalassaemia

SUMMARY A case of double heterozygosity for Aγδβ-thalassaemia and AγGγδβ-thalassaemia was found during a screening programme in Sicily. The proband, a 4-year-old girl, showed a clinical picture of thalassaemia intermedia. Hb F (85–12% by the Singer method) was GγAγ type. The parents and the brother were δβ-thalassaemia carriers. Structural analysis of Hb F showed both Gγ and Aγ chains in the father, but only Aγ chains in the mother.

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