preferable. Of the six cases reviewed by Pagon *et al*,[5] three presented anterior chamber abnormalities with specific reference to Peter's anomaly in one.

As Warburg[7] observes, virtually all multisystem syndromes associated with maldevelopment and non-attachment of the retina are inherited on a recessive basis. Most are autosomal defects although Norrie's disease, in which haemorrhagic detachment is linked with deafness and mental retardation, is an X linked recessive condition.[10] Therefore, especially as consanguinity was a feature of the family described by Chemke *et al*,[4] it would seem that the association of hydrocephalus with congenital ocular abnormalities affecting the retina and, in some instances, the anterior chamber has important implications for genetic counselling.

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Poland-Möbius syndrome

SUMMARY A patient with stigmata of both the Möbius syndrome and the Poland syndrome is presented. This is now the twelfth well-documented patient with a combination of the two syndromes. The association of the Poland syndrome and the Möbius syndrome occurs with sufficient frequency that the combination probably represents a formal genesis malformation syndrome of unknown aetiology that should be designated the Poland-Möbius syndrome.

Both the Möbius syndrome and the Poland syndrome are well reported congenital anomalies. The Poland syndrome consists of absence of the pectoralis major muscle and, in the majority of patients, syndactyly, brachydactyly, and hypoplasia of the hand.[1 2] The Möbius syndrome consists of a variable degree of facial paralysis with inability to abduct the eyes beyond the midpoint.[3] Both syndromes have been associated with a large number of other anomalies. Recently there have been numerous reports describing a combination of the two syndromes in individual patients. It is the purpose of this paper to report a patient who had characteristics of both syndromes and to review briefly the other cases reported.

Case report

This 14-year-old male was admitted to Newington Children's Hospital for bilateral resection of the medial rectus muscles for correction of esotropia. The patient was the 2821 g product of a normal term pregnancy, labour, and delivery. At birth he was noted to have numerous anomalies including

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left hemiatrophy, left talipes equinovarus, absence of the left pectoralis major and minor muscles, a rudimentary left thumb, micrognathia, right calcaneovalgus talipes, bilateral facial nerve paralysis, bilateral abducens palsies, and bilateral dysplastic pinna. He underwent several surgical procedures including removal of the left thumb, cosmetic plastic surgery on his ears, and several procedures on his left foot.

Developmental milestones were normal. There was no family history of congenital birth defects or neurological disease. The parents were not related. Three other sibs are in excellent health.

PHYSICAL EXAMINATION

The patient was co-operative, alert, and of normal intelligence. He had the following findings: bilateral sixth nerve palsies, bilateral peripheral facial nerve palsies, partial ninth and tenth nerve palsies with a weak gag reflex, partial twelfth nerve palsy with bilateral tongue atrophy and weakness, micrognathia, aplasia of the pectoralis major and minor (fig 1), left sided hemiatrophy, left hand and foot acromicria, left hand brachydactyly, cutaneous syndactyly of the left hand, and absence of the left thumb (figs 2, 3). Weight, height, and the remainder of the examination were normal.

RADIOGRAPHIC STUDY

The chest and skull films were normal. *X*-ray of the left forearm revealed underdevelopment of the humerus, radius, and ulna. In the left hand, there was brachydactyly with absence of the first metacarpal and thumb. *X*-ray of the left leg and foot showed underdevelopment of all bones.

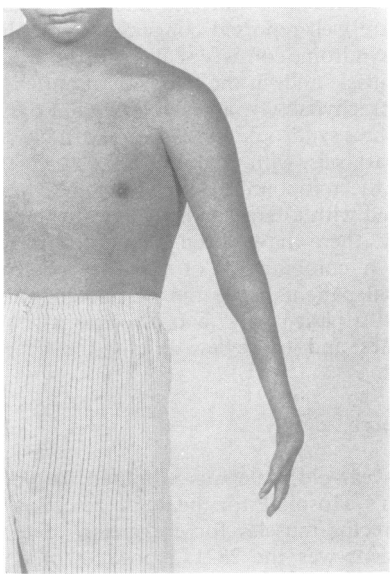

FIG 1 *Shoulder and left upper extremity of patient demonstrating absence of pectoralis muscle and hypoplasia of arm.*

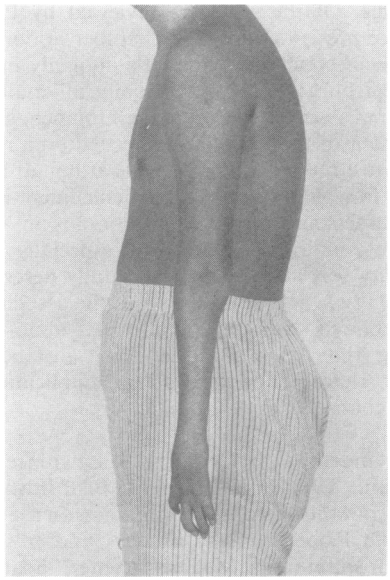

FIG 2 *Left arm of patient demonstrating absence of left pectoralis, left hand acromicria, and brachydactyly.*

CHROMOSOMES

Conventional and G banding analysis showed a normal 46,XY karyotype.

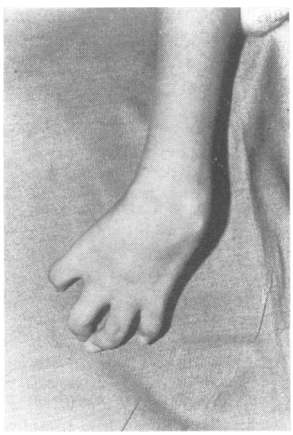

FIG 3 *Left hand of patient demonstrating cutaneous syndactyly, absence of thumb, and brachydactyly.*

Discussion

In 1880, Graefe[4] first described congenital bilateral facial paralysis, and Möbius,[5] whose name has become attached to this syndrome, suggested in 1888 that nuclear agenesis was the pathological lesion. The Poland syndrome was first described in 1841 by Poland[6] who dissected the body of a criminal with unilateral symbrachydactyly associated with ipsilateral aplasia of the sternal head of the pectoralis muscle.

The requirements for inclusion of cases in the Möbius syndrome have varied from author to author. Henderson[7] based the diagnosis on facial diplegia, usually accompanied by palsies of other cranial nerves. Malformations of the limbs were frequently present. Richards[8] required the following as essential features of the Möbius syndrome: unilateral or bilateral loss of abduction of the eyes, unilateral or bilateral facial weakness, and primary or secondary congenital abnormalities of the extremities. Absence of the pectoralis muscles was not unusual. However, no case was presented with the combination of sixth and seventh cranial nerve abnormalities, absent pectoralis major, and symbrachydactyly.

The primary finding in the Poland syndrome is the absence of the pectoralis major muscle. However, the majority of patients have other congenital anomalies. Mace *et al*[2] reported that more than 90% of the 55 patients with the Poland syndrome had syndactyly or brachydactyly or both, along with hypoplasia of the involved hand. David[1] defined the anomaly as congenital unilateral absence of the sternocostal portion of the pectoralis major muscle with ipsilateral syndactyly, a definition also used by Castilla *et al*.[9]

Sugarman and Stark[10] reviewed the published reports and summarised associated malformations in both syndromes. They reported a 5-year-old child with bilateral medial and lateral recti weakness, superior oblique weakness, bilateral facial palsies, and glossopharyngeal and hypoglossal paresis. In addition to these cranial nerve abnormalities, the child had aplasia of the sternal portion of the left pectoralis major, dextrocardia, left-handed acromicria with soft tissue syndactyly, and brachydactyly of the second, third, fourth, and fifth fingers. On radiographic examination, the child had only two phalanges of the index, middle, and ring fingers. They stated that it was the first case reported of a combination of the Möbius syndrome and the Poland syndrome.

Since that paper, other case reports have been reported. Herrmann *et al*[11] in an excellent review of aetiological and phenotypic aspects of orofacial and limb anomalies, reported eight additional patients with the combination of the Möbius syndrome and the Poland syndrome. The authors recommend use of the term Poland-Möbius syndrome to refer to those cases of the 'Möbius syndrome' who have chest muscle defects or symbrachydactyly of the type seen in the Poland syndrome or both. They postulate that these cases of the 'Möbius syndrome', and the majority of cases usually diagnosed as the Poland syndrome, represent a different spectrum of the same condition. Additional combinations of the syndrome have been reported by Szabō[12] and Gadoth *et al*.[13] With the addition of this case, there have now been twelve well-documented cases of the Poland-Möbius syndrome, making this combination more than just a fortuitous circumstance.

The aetiology of the Möbius syndrome and the Poland syndrome remains unresolved. Henderson[7] proposed several theories regarding the aetiology of the Möbius syndrome but concluded that it remains obscure. Thakkar *et al*[14] reviewed the pathology in two cases of the Möbius syndrome and found mineralised necrotic foci in multiple brain stem nuclei and postulated that prenatal encephalomalacia lesions represent the pathological basis for some cases of congenital static Möbius syndrome. Because of difficulties in defining the Möbius syndrome, the role of inheritance in the syndrome also remains unclear. Baraitser[15] reviewed the published cases and concluded that when the definition of the Möbius syndrome includes skeletal malformations, the risk to future offspring is around 2%. The aetiology of the Poland syndrome is also unclear. David[1] noted a high incidence of threatened abortion in children with the Poland syndrome. The association of non-progressive facial, extraocular, lingual, and pectoral muscle agenesis with skeletal deformities probably represents an intrauterine mesodermal defect, as opposed to a self-limited common insult to the fetus, since upper limb budding and cranial nerve nuclei are formed at different stages of fetal development.[13]

The association of the Poland syndrome and the Möbius syndrome occurs frequently enough for the combination to be likely to represent a formal genesis malformation syndrome of unknown aetiology that should be designated henceforth the Poland-Möbius syndrome.

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