CONGENITAL FACIAL DIPLEGIA.

By

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LITERATURE.

Two types of congenital facial paralysis may be recognised. In the first and more common variety the paralysis affects one side of the face only, and is seldom total; nor is it as a rule accompanied by ocular palsies or other congenital malformations. Cases of unilateral facial palsy have been recorded by Stephan¹, Bernhardt², Schultze³, Minor⁴, Marfan and Delille⁵, Souques and Heller⁶, Apert⁷, Goldreich⁸, Falloux⁹, Bonnet-Roy¹⁰, Delherm¹¹, Wilbrand and Saenger¹². In the second type, with which this paper is concerned, the paralysis is bilateral and usually total; in the majority of cases certain of the external muscles of the eye are involved and other congenital malformations may be present. The clinical features of this type appear to have been first described by Graefe¹³ in 1880. His patient was a sane epileptic with facial diplegia (incomplete on the right side), paralysis of both external recti, and total loss of smell and taste. When, as in this case, the paralysis is incomplete, the muscles which escape are almost invariably those in the neighbourhood of the mouth and chin. Of this type are the cases described by Procopovici¹⁴, Möbius¹⁵, Batten¹⁶, Taylor¹⁷, Fry¹⁸, and Woltmann¹⁹.

Complete paralysis of the sixth and seventh pairs has been reported by Harlan²⁰, Möbius²¹, Decroly²², Fryer²³, Schapring²⁴, Kahlmeter²⁵, Kirby²⁶, Chisolm²⁷, Cabannes²⁸, and Leszynsky²⁹. The anatomical relationship of these nerves within the brainstem explains why they are so frequently implicated together, but it is also possible for the third and fourth cranial nerves to be involved, as in Recken's³⁰ case, where partial facial diplegia was accompanied by a complete external ophthalmoplegia, or again as in a case reported by Möbius³¹ where the inferior recti were paralysed.

Less frequently three or four cranial nerves are concerned. Thus, paralysis of the motor fifth has been recorded by Schapring²⁴, and by Fry and Kassak³²; paralysis of the facial and hypoglossal nerves by Woltmann¹⁹ and Schmidt³³; and paralysis of the sixth, seventh and twelfth by Kirby²⁶ and Batten¹⁶. With the solitary exception of the case of Graefe, referred to above, there are no records of disturbance of the sensory cranial nerves, and it is important to note that although the extrinsic ocular muscles are often affected those concerned with pupillary reactions invariably escape.

The congenital malformations which may be encountered are very diverse. Double epicanthus has been described by Schrapinger²⁴, bilateral absence of

the lachrymal caruncles by Schrapinger²⁴ and Chisolm²⁷, deformed ears by Thomas³⁴, funnel breast by Schrapinger, absence of the pectoralis major and mamma by Woltmann¹⁹, prominence of the eyes by Thomas³⁴, recession of one eye by Graefe¹³, fused or dwarfed fingers by Möbius¹⁵, Saignon and Woltmann¹⁹, shortness of limbs by Fryer²³, club foot by Kirby²⁶ and Lennon²⁹ and diastasis of the abdominal muscles by Lennon. Saignon and Bertein³⁵ refer to the possibility of visceral lesions.

SEMIOLOGY.

In a typical case the appearance of the patient is most striking. The face is sunken and mask-like, without lines or expression; the lips, full and protruding, are parted so that saliva may dribble from the mouth; the skin covering the atrophied facial muscles is smooth, shiny and pale. When the patient attempts to close the eyes the globes move upwards and a little outwards or inwards. If to the above picture there is added paralysis of the sixth cranial nerves, movement of the eyes to either side is impossible, and there is usually a marked degree of internal strabismus. In spite of this, convergence may be retained. Only exceptionally is acuteness of vision impaired, as in the cases reported by Harlan²⁰ and Möbius¹⁵.

Examination of the electrical reactions is of great importance, for in complete facial diplegia of congenital origin both muscles and nerve invariably show an entire absence of response to either galvanism or faradism. Occasionally certain muscles, especially the platysma and those of the chin, retain their excitability, but the reaction of degeneration is never found.

Owing to inability to approximate the lips the patient has difficulty in pronouncing the consonants b, f, m, p and v.

ETIOLOGY AND PATHOLOGY.

Unilateral facial paralysis is not infrequently the result of injudicious use of forceps at birth, but bilateral obstetrical paralysis must be of exceptional rarity, for Edgeworth³⁶ appears to be the only writer to have recorded a case in recent years. Nearly all cases of congenital facial diplegia are attributable either to an agenesis or to a degeneration of the cells of the facial nucleus: only in one or two has the defect been in the mesoderm. Lennon²⁷, for example, refers to a case reported by Schenkl in which the facial muscles were absent, although the nerves themselves and their central connections were intact. Möbius's²¹ suggestion that the condition owes its origin to some defect of the facial nucleus is very largely born out by the few pathological investigations available in the literature. Heubner³⁸ performed an autopsy on a child, age two and a half years, who during life presented total left and partial right facial paralysis, paralysis of both external recti and atrophy of the left half of the tongue. Histological examination showed atrophy of the cells of the sixth, seventh and twelfth cranial nerves, with, in addition, imperfect development of the left olive and left pyramidal tract. Sections stained by

Marchi's method showed no evidence of recent myelin degeneration ; examination of the facial nerves was confined to their intramedullary portions. Heubner concluded that there was a hypoplasia of the nuclei concerned, but this view is decidedly open to question, for the mere absence of nerve-cells is no proof of nuclear agenesis. In old-standing lesions it is never easy to distinguish between aplasia and atrophy, and Heubner's description of the pathological changes would apply equally well to a degeneration or atrophy of nerve-cells.

In another case, reported by Rainy and Fowler³⁹, the lesions were of recent origin and their investigation yielded important information. An infant ten weeks old was admitted with almost complete paralysis of both facial nerves. Eye movements were normal and no other cranial nerves were affected. At first the orbiculares palpebrarum responded to galvanic and faradic currents, but very soon these reactions faded out, without the occurrence of R.D. The child succumbed to broncho-pneumonia. Examination of the brainstem showed disappearance of many of the nerve-cells of the facial nuclei, those remaining showing shrinkage, irregular Nissl bodies and ill-developed processes. The sixth nuclei were intact. Marchi's stain revealed degeneration of the fibres of the facial nerves throughout their whole course ; the facial muscles which the authors described as being greatly atrophied proved normal on microscopic examination.

In this case, since degenerated nerve-fibres were present, it is obvious that at an earlier stage there must have been nerve-cells from which they took origin, and consequently aplasia* as a possible cause could be excluded. The authors remark that since the electrical reactions appeared to fade out without the occurrence of R.D. "one must conclude that no sudden accident whether of the nature of a traumatism or an acute polioencephalitis abruptly severed the muscles from their nutrient centres, while the bilateral symmetry of the lesion is also adverse to such an opinion." Concerning the nature of the toxin or other agent which could cause nuclear lesions limited so strictly that while the nerve cells of the seventh nucleus were profoundly affected those of the sixth remained normal they have nothing to say save that "the nerve cells slowly perished, leaving evidence of their former existence in the degenerated nerves and in the atrophied muscles they formerly controlled."

Were it not for the absence in this case of R.D. one would be tempted to regard their case as an example of traumatic facial paralysis, for since marks caused by the obstetrician's forceps were plainly visible five weeks after birth the original trauma must have been fairly severe. Moreover, the histological appearances were quite compatible with those which follow an indirect Wallerian degeneration such as might have arisen after bruising or crushing of the facial nerves.

REPORT OF A CASE.

V. S., a male imbecile, age 8, was admitted to hospital on February 22, 1909. No history could be obtained. He was noted to have strabismus,

club-foot and undescended testicles, but the facial diplegia escaped detection and was not recognised until nearly twenty years later.

State on Examination: April 1, 1929. The patient is an undersized adult male Jew. Height 61 inches. The upper limbs are disproportionately long and the lower limbs short; the latter show a slight degree of genu valgum and bilateral talipes varus. There is hair on the chest, sacrum and lower limbs; on the latter situation the growth is remarkably abundant. The heart and lungs are healthy and the Wassermann reaction in the blood is negative. The head is small with sloping forehead and occiput: circumference 21 inches.

The facial paralysis is complete and total, neither voluntary or emotional movement being possible. The forehead is high, receding and entirely free from wrinkles. The face is expressionless, the complexion pale and the cheeks hollow, especially on the left side. The palpebral apertures are wide and both upper lids are retracted so that the eyelashes are hidden at their bases. The eyebrows, which are situated a few millimetres above the free edges of the eyelids, are thick and straight. Winking does not occur.

On both sides a marked degree of internal strabismus is present and it is perhaps a little more pronounced on the left. The eyes cannot be moved outwards but a slight degree of convergence is possible. All other ocular movements are performed fairly well. When the patient attempts to close his eyes the globes move upwards and inwards. Slight narrowing of the palpebral apertures accompanies the upward movement of the eyes but this appears to be due not to any contraction of the orbiculares but to a passive lifting of the lower lids through the agency of the conjunctiva.

The pupils are equal and react normally to light. Vision is remarkably good; in reading the patient turns his head obliquely to one side.

The nose is somewhat bulbous, the nares are narrow, and on deep inspiration the alae nasi are indrawn. The ears are small, symmetrical, and well formed; the mastoid processes are normal and there is no impairment of hearing. The mouth is habitually open and saliva occasionally dribbles from the everted lower lip. Upward and downward movements of the lower jaw are performed freely without deviation but lateral movements are almost nil.

The patient is quite unable to bring his lips together and has therefore to adopt special devices for dealing with liquid food or when smoking. When, for example, soup has to be swallowed he tilts his head well back so as to retain it in his mouth. In order to smoke a cigarette he covers his mouth with one hand and holds the end of the cigarette between the third and fourth digits. Smoke can be readily expelled through the nose even when his mouth is uncovered. The tongue is protruded straight and shows neither atrophy nor tremor; its tip, however, is unusually pointed. The soft palate and uvula move normally on phonation.

The voice is deep and guttural, and the patient sings in a low monotonous tone; the complete absence of facial movement during his recital reminds one of the performance of a ventriloquist.

The skin of the face is unusually thin and pale, and when the underlying tissues are pinched up between the thumb and forefinger the absence of muscular development is very obvious. Sweating on this area may occasionally be noted, and there is a vigorous growth of hair, necessitating frequent shaving.

All forms of sensation are normal.

Electrical reactions : There is an entire absence of response to either the faradic or galvanic current in the facial nerves and muscles. The patient's mental state is one of medium grade imbecility. Mental age, 7 years.

DISCUSSION.

Although no history is available there can be little doubt that the facial diplegia in this case is congenital. The bilateral character of the paralysis, its association with sixth nerve palsy, the absence of contracture, and the occurrence of faults in development, such as club-foot, all point in this direction, and to this view the complete absence of electrical reactions lends additional support.

Further, it must be noted that in addition to paralysis of the sixth and seventh cranial nerves there is involvement of part of the motor division of the fifth, and in this respect the case bears a resemblance to one of Schapringer's cases. Finally, it may be noted that this appears to be the first case in which an obvious degree of mental defect has been present.

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Fig. 1. Normal expression.

Fig. 2. Attempt to 'screw up' eyes.

Fig. 3. Profile.

Fig. 4. Bilateral club foot.