CONGENITAL BILATERAL TOTAL EXTERNAL OPHTHALMOPLEGIA COINCIDENT WITH OTHER DEVELOPMENTAL ABNORMALITIES

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The following case is considered worthy of report as it presents an unusual combination of congenital abnormalities.

CASE REPORT

S.F., a Coloured female aged 17 years, was admitted to Baragwanath Hospital on 7 August 1956. According to her mother she had been unable to open her eyes properly since birth; she could not see well with her right eye, but vision in the left eye was normal. The mother had never noticed any squint, nor had the patient complained of diplopia. It was further stated that the patient had been born with a protruding chest. Her milestones with regard to mental development were normal. Menstruation started at the age of 15 years and was normal. There was no history of past illness.

The patient was the last of 6 siblings and was born after a normal pregnancy and labour. There was no history of congenital abnormalities in other members of the family. We examined 40 immediate relatives and found no obvious congenital abnormalities.

Examination

The patient was of normal intelligence and showed no disorder of affect, behaviour or speech. Her measurements were: height 59 inches, span 66 inches, floor to pubis 32 inches. She stood with her head thrown back, the sternomastoid and trapezius muscles prominent. The dorsal spine was the seat of a marked lordosis in its middle portion and was associated with forward protrusion of the thorax (Fig. 1). The cervical spine was normally mobile but the rest of the vertebral column was rigidly fixed. Her gait was normal and her agility surprising in view of the severity of the spinal deformity. The lobes of both ears were vestigial and were joined to the neck by a fold of skin.

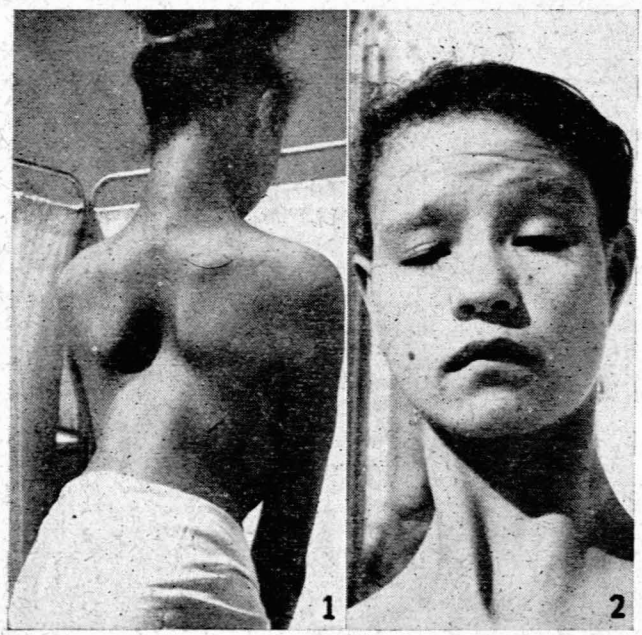

Fig. 1. The lordotic deformity of the thoracic spine.
Fig. 2. The bilateral ptosis, compensatory contraction of occipito-frontalis muscle and prominent sterno-mastoid muscles.

Bilateral ptosis was present, reducing the palpebral fissures to mere slits. Compensatory contraction of the frontalis muscle was well in evidence. The eyeballs were fixed in the straight-forward position, no movement being possible in any direction (Fig. 2). There was no response to 1·5 mg. prostigmine by intramuscular injection. The pupils were round and equal, reacting to light but failing to contract on accommodation. Visual acuity of the left eye was normal, while that of the right eye was poor (6/36 Snellen's types). The visual fields and colour vision were normal. On retinoscopy the patient was astigmatic and myopic. The right optic disc was transversely elliptical and appeared to be enlarged. The lateral half of the disc was irregularly pigmented with numerous small blood vessels ramifying over its surface. Fundoscopy of the left eye was normal. The remaining cranial nerves were normal, as was the rest of the central nervous system on examination. Pubic and axillary hair were scanty but breast development was satisfactory. Examination of the heart, lungs, abdomen and urine was normal.

Radiological Findings (Dr. A. Berezowski)

Skull: The vault is rather small and presents a beaten silver appearance probably consistent with the patient's age. The pituitary fossa is normal in size and no intracranial calcification is seen. Platybasia is noted.

Cervical Spine: There is a spine bifida of C4 and fusion of the posterior elements of C5 and C6. The appearances are consistent with a Klippel-Feil type of abnormality.

Dorso-lumbar Spine (Fig. 3): Most of the bodies are rather longer than broader. There is fusion of the rib elements posteriorly as well as of most of the dorsal elements of the vertebral bodies in the thoracic region. This has resulted in a marked lordosis of the mid-dorsal spine. The vertebral bodies in this region are very small.

Lumbar Spine: There is rotational scoliosis convex to the right. An opacity, 0·5 cm. in diameter, lying between D12 and L1 on the left side probably represents an unusual ossification centre.

Pelvis: There is some asymmetry caused by maldevelopment of the left hemipelvis.

Laboratory Investigations

Haemoglobin 14·8 g.%. Paper electrophoresis revealed no abnormal haemoglobins. Leucocyte and differential counts were normal. Blood group O, Rh-positive, cDe, MNS. Standard Eagle test negative. The blood urea and carbon-dioxide-combining power normal; plasma chloride, cholesterol, inorganic phosphorus normal; serum sodium, potassium, calcium and bilirubin normal. Alkaline phosphatase 6·4 units (King-Armstrong). None of the following substances were detected in the urine: pentose or other reducing substances, porphyrins, homogentisic acid, amino acids in excess. pH of urine 6·3.

DISCUSSION

Giri[1] has summarized the main features of congenital paralyses of the ocular muscles as follows: 'Ptosis is the com-

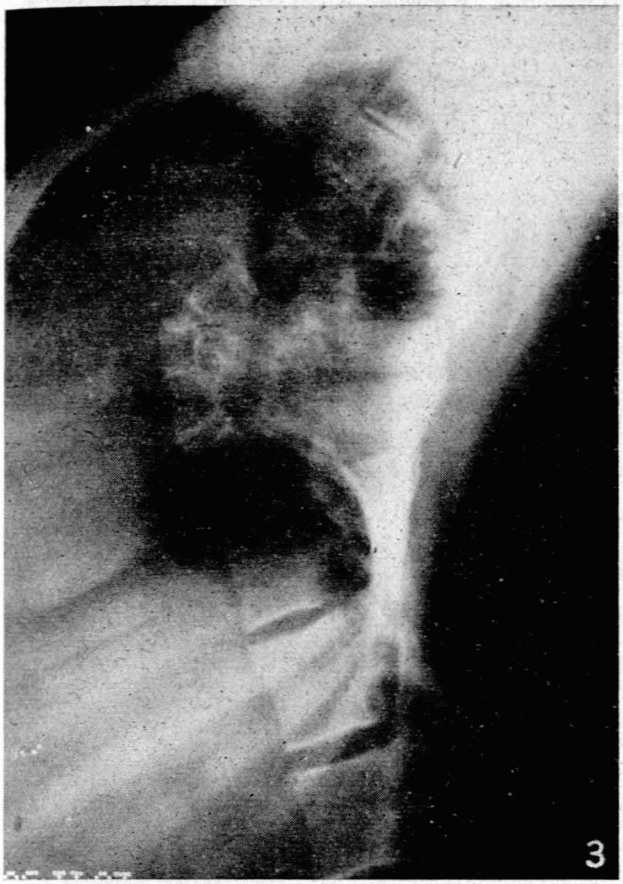

Fig. 3. X-ray of dorso-lumbar spine showing the lordosis and abnormalities of the vertebrae.

monest. Next come paralyses of one or more muscles either in conjunction with or independently of ptosis, affecting the two eyes similarly or differently, some of the muscles having preserved their function either partially or completely. The affection may be unilateral or bilateral. Divergent or convergent squint is often present. Errors of refraction have been noted in most cases. Almost total bilateral ophthalmoplegia externa with ptosis appears to be the least common'.

Complete bilateral ophthalmoplegia externa occurs sporadically or may be inherited either as a Mendelian dominant[2] or as a sex-linked recessive.[3] The theories advanced to explain the anomaly have invoked either a nuclear or a muscular defect. Moebius[4] attributed the condition to aplasia of the nuclei in the brain stem while Langdon and Cadwalader[5] reported a diminution in the number and shrinking of the cells of all the oculomotor nuclei except the Edinger-Westphal nuclei. Uthoff[6] suggested that a congenital defect in the anlage of the ocular muscles was responsible for the anomaly. Fuchs,[7] on the basis of biopsy studies, believed that the condition represented a myopathy of the extra-ocular muscles.

In the case described above the following points call for comment. The failure of the pupils to contract on accommodation has been attributed to myopia and the inability to converge.[3] Both the pigmentation of the optic disc and the vascular malformation of the retinal vessels are developmental defects which may cause visual impairment[8] and probably explain the poor vision in the patient's right eye.

This case was remarkable for the multiplicity of developmental abnormalities involving the axial skeleton. Thus there was a platybasia, a Klippel-Feil deformity of the cervical vertebrae, asymmetry of the pelvis and a dysplasia which resulted in a severe lordosis of the dorsal spine and the total loss of mobility of the vertebral column below the cervical region. The association of congenital bilateral total external ophthalmoplegia with other developmental anomalies, such as spina bifida and cardiac and thoracic malformations, was also noted by Salleras and de Zarate in their sex-linked recessive cases.[3]

SUMMARY

A case is reported of congenital bilateral total external ophthalmoplegia associated with other developmental anomalies involving particularly the axial skeleton.

We wish to thank the Medical Superintendent of Baragwanath Hospital for permission to publish this case, Dr. K. Keeley for his advice on the investigation of the case, Dr. J. Rom for the ophthalmological examination and Dr. S. Kimmel and Mr. A. Shevitz for the photographs.

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