SURGICAL TREATMENT OF THE KLIPPEL-FEIL SYNDROME

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Though we must go back to Herodotus for the earliest account of the mythical race of Acephala (Fig. 1), who were supposed to inhabit the western part of Lybia, we owe to Ulisses Aldrovandi (1522-1605) the suggestion that the "monstrosity" consisted simply of absence of the neck, the head and chest being so closely approximated that the eyes seemed to belong to the chest (Belloni 1950). There is no doubt that the ancient inventors of the myth had seen an example of severe congenital deformity of the cervical spine.

Haller (1743) and Morgagni (1746) described the anatomical features in two such cases with reduction in number and fusion of the vertebrae. But it was not until 1912 that an adequate clinical and anatomical description of this anomaly was given by Klippel and Feil, who regarded the deformity—perhaps wrongly—as a single well defined entity.

FIG. 1
The Acephalus. A sculpture by Nicholaus in the Cathedral of Ferrara (Italy).

The Klippel-Feil syndrome, or congenital osseous torticollis, was studied later by numerous authors, and especially by Bertolotti (1920), who recognised the following essential features: 1) absence or shortening of the neck; 2) limitation or absence of movements of the neck; and 3) a low hair line at the back of the neck (Fig. 2). There might also be other minor features, such as thoracic and cranio-facial asymmetry, flat back, simian deformity of the scapulae, impaired jaw movement, angular cervico-thoracic scoliosis, and occipital platycephaly; and the condition might be associated with congenital deformities in other parts of the body.

According to Bertolotti the anatomical changes seen radiographically include fusion of the vertebrae into one mass, either cervical or cervico-thoracic, hemivertebra, platyspondyly, atlanto-occipital fusion, assimilation of the third cervical vertebra with the axis, transverse segmentation of vertebral bodies and vertical segmentation of bodies and arches, basal segmentation of the odontoid process, partial absence of the sacrum, and total absence of the coccyx.

These diverse manifestations lead me to believe that the Klippel-Feil syndrome should not be regarded as a single clearly defined entity, but merely as a condition of cervical deformity with or without other malformations.

Hitherto the accepted practice has been either to ignore the problem of treatment or to treat the condition symptomatically. With the advances that have been made in surgery I believe that in severe cases the policy of non-intervention should be discarded and an attempt made to correct the cervical deformity with the object of improving both appearance and function. I shall describe the surgical treatment of the Klippel-Feil syndrome under four headings: 1) operations on skin; 2) operations on muscle; 3) operations on nerves; and 4) operations on bone.

OPERATIONS ON SKIN

The Klippel-Feil syndrome is often encountered in the "status" of Bonnevie Ullrich (Turner syndrome), the most striking feature of which is the "pterygium colli" consisting of asymmetrical creases extending from the mastoid process to the acromion, and sometimes involving the muscles and fasciae as well as the skin. Though neck deformities are often present the sex incidence distinguishes this syndrome, which occurs mainly in girls, from

other varieties of the Klippel-Feil syndrome, which are commonest in boys. The three cases of pterygium colli (all in females) that I have studied from the records of Professor Sanvenero Rosselli were all associated with deficiencies of the cervical or thoracic vertebrae (Fig. 3).

FIG. 2

A patient with typical Klippel-Feil disease (*right*) compared with a normal girl of the same age.

The operation of double z-plasty, with resection of part of the muscle or fascia when necessary, afforded permanent correction of the deformity and allowed greater freedom of neck movement, though movement was still restricted by the bone deformities.

OPERATIONS ON MUSCLE

In some of these cases there is a bilateral contracture of the sternomastoid muscle which is amenable to correction by surgery. In one case, in which I used Putti's technique, the posture and mobility of the neck were much improved. Through a sub-mastoid incision (the scar is hidden by the ear) the mastoid insertion of the sternomastoid muscle is dissected from the bone and a segment three centimetres long is removed. The clavicular and sternal heads of the muscles are divided by subcutaneous tenotomy to avoid leaving scars. Correction is maintained at first by a Schanz collar and later by a plaster case applied with over-correction and retained for two or three months. The correction obtained by this method might well be further improved by stretching or division of other muscles, such as platysma and trapezius, which are frequently also contracted.

FIG. 3

An example of pterygium colli (syndrome of Bonnevie Ullrich) with congenital deformity of the cervical spine before and after plastic surgery (case of Dr Sanvenero Rosselli).

OPERATIONS ON NERVES

Disorders of the spinal cord and compression of nerve roots have been described in the Klippel-Feil syndrome and they doubtless have the same congenital origin. Spasmo-cerebellar syndromes (Serafini and Bertolotti), spastic hemiparesis (Sicard and Sermayez), spastic pyramidal quadriplegia (Guillamme and Mollaret), all with late onset, are examples. Brachial pain is also frequent, as well as painful trophic syndromes of the brachial plexus beginning after the twentieth year of life. Such cases have not so far been treated surgically.

These radicular syndromes, surely connected with the existence of cervical ribs, hidden bifid spines, or congenital deformities of either the radicular or plexus distribution, might certainly be improved by operative removal of supernumerary ribs or by hemilaminectomy or foraminotomy.

It is in fact a notion now commonly accepted, despite its novelty, that congenital deformities affecting the position or distribution of nerve roots can be found in the sacral plexus which may be responsible for severe lumbar pain and sciatica, whether or not they are

FIG. 4

Klippel-Feil disease associated with left congenital high scapula. The upper photographs show the child at the age of four, before operation. The lower pictures show the child aged nine, five years after operative lowering and fixation of the left scapula.

associated with bony anomalies (spina bifida), or with disturbance of the ligaments or intervertebral discs; and these disorders have been treated successfully by operation.

The same decompressive surgery could be attempted also in the cervical spine; but there the risk and difficulty would be greater.

Although many authors have given a detailed description of the pathological bony

anatomy in the Klippel-Feil syndrome the associated congenital disturbances of the spinal cord have surprisingly been ignored, as have the anomalies in the distribution of the nerve roots which are certainly associated with the deformity in many cases. Anatomical investigations in this direction should bring important results, as they have in disorders of the lumbo-sacral region of the spine.

FIG. 5

Same patient as shown in Figure 4. Radiographs before and after operation.

OPERATIONS ON BONES

Various congenital deformities of the shoulder girdle are often associated with the Klippel-Feil syndrome, among which the congenital high scapula is common. In fact this condition is nearly always associated with deformities, however slight, of the spinal column, and, especially when the scapula is fixed to the cervico-thoracic vertebrae through a bony bridge having the form of a rib, it may be responsible for severe deformities with limitation

of movement of the neck and upper limb (Figs. 4 to 5). Operations that I have undertaken for the relief of congenital high scapula have followed Putti's technique, which has always given satisfactory results consistent with the degree of deformity, the age of the patient, and the complications that are sometimes present in severe cases, such as paresis of the brachial plexus. The steps of the operation may be summarised as follows. The incision curves along the vertebral margin of the scapula. The lower part of the trapezius and the upper part of the latissimus dorsi are divided. Below the rhomboid muscle the bony rib-like process that anchors the scapula to the cervico-thoracic vertebrae is isolated and removed. The scapula is freed and severed from its fibro-muscular attachment. It is brought down until its lower angle is fixed to the fifth rib. The reduction of the scapula is maintained by a plaster shoulder spica for some weeks before movements are begun. In one patient operated upon by Putti in 1908 the clavicle also had to be divided before adequate function of the scapula could be secured.

Osteotomy of the spine as described by Smith-Petersen *et al.* (1945) and La Chapelle (1934) and by Delitala and Pais (1952) for the correction of deformities at sites involving less risk could be carried out with extreme caution also for bony torticollis and for the typical cervico-thoracic flexion deformities which, according to some authors, can simulate the late results of spondylitis. Such an operation would sometimes be practicable because the intersomatic joints are often preserved and interrupt the cervico-thoracic synostosis. A cervical osteotomy was in fact carried out successfully by Herbert (1948) and another was undertaken by Law (1951) but the result was poor.

Since in most cases an operation on the cervico-thoracic block is technically impossible I studied the possibility of obtaining a "cervicalisation" of the first thoracic segment by means of bilateral upper thoracic thoracoplasty, with the idea not only of correcting angular deformity but also of forming a new neck. In many examples of the Klippel-Feil syndrome—and always in the more severe cases—the first four ribs are found much above the level of the shoulder girdle, thus giving the typical appearance that has been termed "cervical chest." There is also a high incidence of unilateral or bilateral cervical rib in these cases, as well as of other minor variations such as low implantation of the breasts. The operation that I have devised therefore seems perfectly rational. I was unable to find record of an operation of this nature ever having been tried before. Nevertheless I believed that removal of the upper ribs, which are often supernumerary or congenitally interlinked through synostoses, would not seriously impair respiration, and experience of pulmonary surgery for tuberculosis lent support to this view.

CASE REPORT

The patient was a woman of thirty-two with severe Klippel-Feil syndrome of the classical type. Radiographs showed slight platybasia. The posterior arch of the atlas was assimilated in the occiput, and the body of the atlas was no more than a vestige, scarcely differentiated from the posterior arch. The other cervical vertebrae were fused into a solid block. Bilateral cervical ribs were present, the left one seemingly fused with the first rib. There was a rudimentary cervico-thoracic hemivertebra which appeared to be the cause of a high thoracic scoliosis convex to the right. The patient was able to rotate the head through a few degrees, but all other movements were abolished.

Operation—Partial thoracoplasty was carried out on the right side on May 10, 1954, under local anaesthesia (Fig. 6). Through a right paravertebral incision midway between the spinous processes and the medial margin of the scapula, the trapezius and rhomboid muscles were divided, exposing the first four ribs. The posterior arches of the uppermost three ribs were excised, beginning with the third. The first rib was removed together with the supernumerary cervical rib with which it was fused. The ribs were not disarticulated, but were divided a few centimetres from the joint in order to save time and to avoid excessive lowering of the apex of the lung, which is liable to be anchored to the posterior costo-vertebral angle. The wound was closed completely in layers.

Progress—The post-operative course was uneventful. Immediately after the operation there was a noticeable improvement in the shape of the neck on the right side, and the patient was able to flex and extend the neck for the first time. Radiographs a few days after the operation showed a subcutaneous air-bubble and a haematoma above the lowered dome of the pleura,

FIG. 6

Diagrams illustrating the operation of right partial thoracoplasty of the seventh cervical and first, second and third thoracic ribs. The diagrams on the right show tracings from radiographs of the neck (a) before and (b) after right partial thoracoplasty. The diagrams below show the dimensions of the costal segments removed. The seventh cervical and the first thoracic ribs were fused into one large rib 8 centimetres long and 4 centimetres broad. Six months later the same operation was performed on the left side.

evidence that pulmonary function was not damaged. Two weeks after operation a plastic collar was applied to mould the regenerating ribs to the improved contour.

Four months after the first operation the second stage was carried out successfully on the left side, the first, second and third ribs being resected under local anaesthesia.

The patient was discharged from hospital ten days after the second operation and after a further month she was able to return to work. In the meantime she was treated by physiotherapy, with massage and daily cervical suspension for half an hour.

FIG. 7

Radiograph of the upper thorax before operation.

FIG. 8

Radiograph of the upper thorax after bilateral partial thoracoplasty.

At follow-up examination four months after the second operation the patient's general condition was good. She had gained weight, and did not complain of any difficulty with respiration. Spirometry was 1,060 cubic centimetres, scarcely lower than before operation. Rotation, flexion-extension, and lateral flexion at the cervico-thoracic junction allowed good

FIG. 9

The patient before and after operation.

mobility, a little less than the normal. The scoliosis was also improved. Radiographs showed that with the lowering of the pulmonary apices and the disappearance of the "cervical thorax" the uppermost three thoracic vertebrae, freed from the ribs, had acquired the appearance and function of cervical vertebrae (Figs. 7 and 8). Traces of periosteal ossification on the right did not affect the shape of the new neck (Figs. 9 and 10).

FIG. 10
Back view of the patient before and after operation.

Comment—I believe that the method of treatment described here is free from serious risk if performed with a careful technique. It provides appreciable correction of an ugly congenital deformity which was hitherto considered unsuitable for surgical treatment. A larger experience of the method, though not easily obtained because of the rarity of these cases, will probably bear out these optimistic conclusions.

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