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An uncommon malformation simulating a duplication of the spine

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As a further contribution to our recent studies in collaboration with Professor Luigi Gedda on the vertebral deformities and their relation to the « schysosynostosis axialis congenita familiaris », we believe of interest to report here a case of quite unusual congenital cleft of the lumbar spine, associated with other deformities of the axial skeleton.

Congenital clefts due to developmental anomalies may occur either in the neural arch or in the vertebral body. The former are usually called « spina bifida » and in their simplest form, i.e. « spina bifida occulta », are very common at the lumbosacral level in asymptomatic individuals. The fissure of the vertebral body, also known as « butterfly-vertebra » or « somatoschisis » or « spina bifida anterior », is much less common: in an a.p. radiogram it appears as a sagittal clear space not more than a few millimeters in width, dividing the vertebral body into two halves; usually, the corresponding neural arch is complete and only one or sometimes two adjacent vertebral bodies are affected.

Even less frequent are the cases of complete gap of both the bodies and the neural arches of several adjacent vertebrae, accompanied by wide separation of the two halves, so as to simulate a duplication of the spine. Such cases have been occasionally reported in dead fetuses, usually in association with other malformations not compatible with life. Instances of this anomaly appear to be extremely rare in the adults (29). In this paper a case will be reported of wide disruption of the lumbar spine observed in an otherwise healthy woman.

Case report

The patient is a 25 year old woman. She is of unknown parents and therefore familial data are not available and past personal history is vague and incomplete. According to her own informations, the patient has not suffered from important diseases and has never experienced any trauma. Since her first years of life the left shoulder appeared to be more elevated than the right and the movements of the trunk considerably impaired. It is noteworthy, howewer, that the patient has been always able to work as a housemaid without significant impediment.

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She has had regular menstruations since her 13th year. In 1955 she delivered without unusual difficulty a healthy male baby born from an illegitimate union. The baby is now 6 months old and is free from any malformation, both clinically and radiographically.

Physical examination (Fig. 1a and 1b).

Stature 158 cm., weight 61 Kg.

The head is slightly tilted to the left and the face is somewhat asymmetric with sagging of the left angle of the mouth. Oral cavity and teeth show no abnormalities.

The neck presents an evident flattening of the normal lordosis, and there is some limitation in the backward movement and lateral bending; flexion and rotary motion are not impaired. The nuchal hair line is normally situated.

The trunk is short and stocky, so that the lower ribs almost reach the iliac crests. The left scapula is elevated and therefore the upper portion of the trunk is grossly asymmetric. There is scoliosis to the left in the cervicodorsal region and scoliosis to the right in the midthoracic area. The dorsal kyphosis is flattened, whereas the thoracolumbar region presents an angular gibbus (fig. 1 b). At the same level, on the midline, there is an irregular bumpy protuberance, showing several umbilications and overlaid by normal skin (fig. 1 a). On palpation the mass appears to be soft, its consistency resembling that of a lipoma, and is delimited by multiple osseous spurs. Throughout the area corresponding to the mass one fails to palpate the spinous processes of the vertebrae on the midline; a deeper pressure gives rise to local pain.

The movements of the thoracic spine are markedly limited, and those of the lumbar column are practically abolished at all.

The pelvis is asymmetric, the right half being larger than the left. The abdomen is prominent, because of the shortening of the trunk.

The gait is somewhat impaired, owing to derangement of the statics determined by the asymmetry of the trunk and pelvis. Active and passive movements of upper and lower extremities are normal and cause no painful sensations. The right leg is slightly hypotonic and hypotrophic. A large zone of hypesthesia can be elicited on the anterior surface of the right thigh, corresponding to the innervation area of the second and third lumbar roots. In the same part of the thigh the patient has often complained of loss of sensation and numbness. The deep reflexes are diminished at the right lower limb.

Bowel and bladder functions are referred to be normal.

Other details of the physical examination and laboratory data are without interest for the present article.

Roentgen examination.

Skull. The vault and the base appear to be normal. There is no sign of basilar impression.

Cervical spine. There is an evident flattening of the cervical lordosis. The articulation between the atlas and the occipital condyles is normal, and so are the succeeding five intervertebral spaces. The disc between C_6 and C_7 is incomplete, and the bodies of the two vertebrae are partially fused together. The neural arch of C_6 shows a sagittal cleft. The lower cervical spine presents a slight scoliosis to the left (fig. 2).

Dorsal spine (fig. 2). D_1 and D_2 are normal. The bodies of D_3-D_5 are fused into a wedge-shaped bony block with its basis on the left side and poor evidence of remnants of the intervertebral discs: at this level the spine is markedly scoliotic to the right. On the right side the ribs corresponding to this osseous block are partially fused near their attachment to the spine; on the left they are arched cephalad, causing an image resembling spider claws.

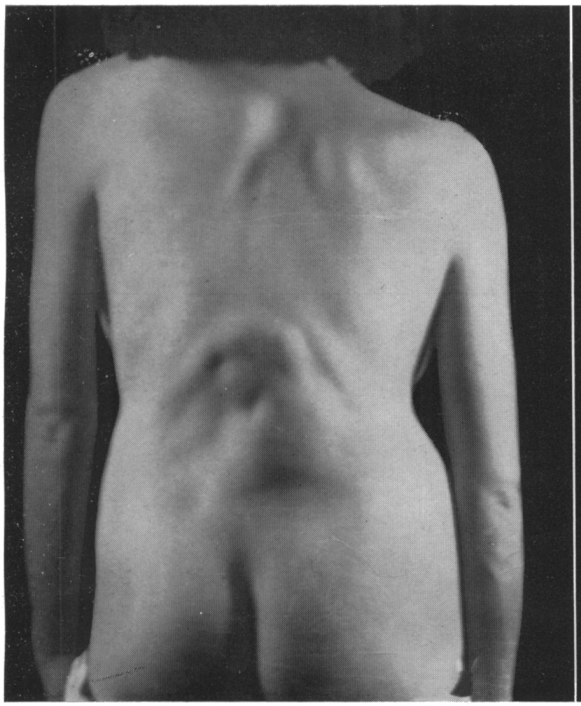

Fig. 1a

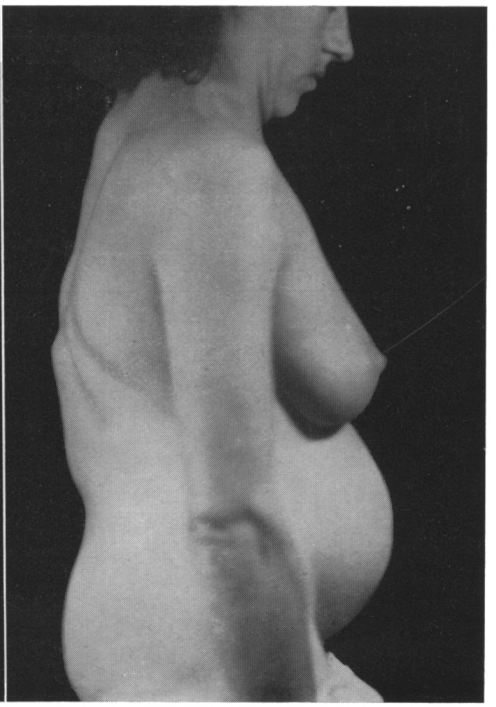

Fig. 1b

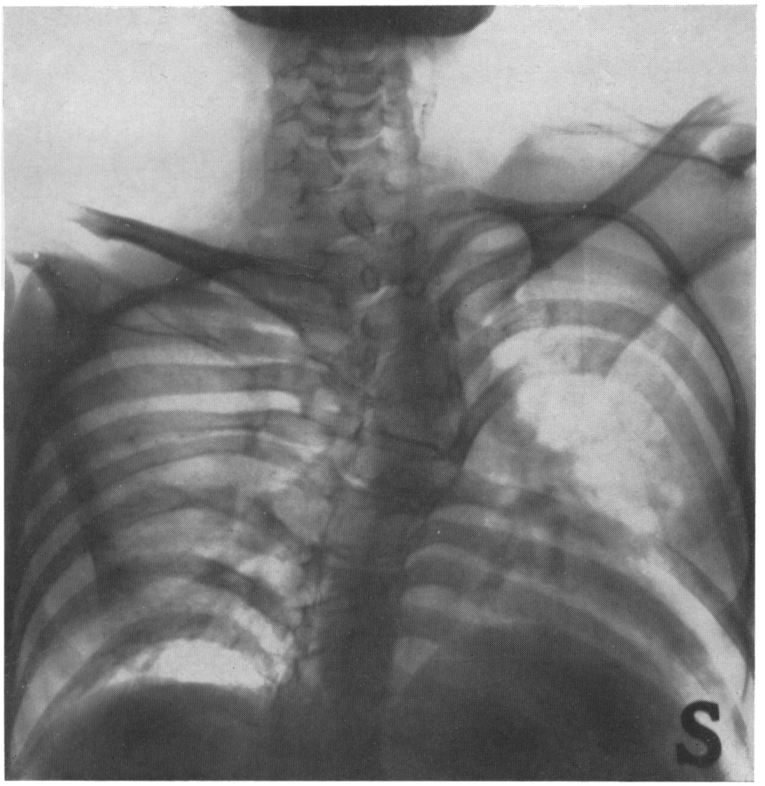

Fig. 2

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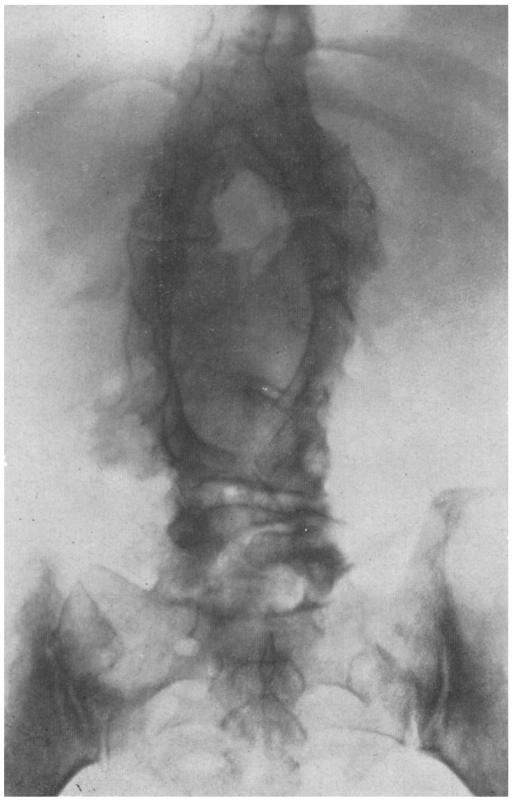

Fig. 3a

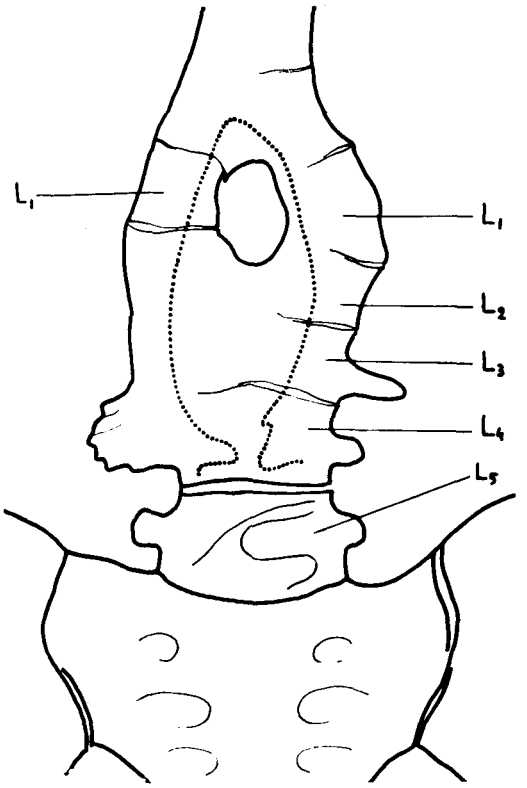

Fig. 3b

D_6 is obliquely placed, but does not show any other abnormality; the neighbouring intervertebral spaces are free. The following thoracic segments are partially fused into another vertebral block in which the individual vertebrae are hardly discernible. This thoracic block is divided in its distal portion assuming a fork shape, which continues in the gross lumbar deformities to be described below (fig. 3a).

The total number of the ribs is reduced to eleven on each side.

The lateral projection shows a flattening of the kyphotic curvature of the upper thoracic spine. At the dorso-lumbar junction there is an angular kyphosis; at the fulcrum of the angulation a small dorsal hemivertebra is present, probably corresponding to D_{12}. Intervertebral foramina of the lowest thoracic segments appear to be somewhat misshaped.

Lumbar spine. This portion of the vertebral column shows the most extensive deformities. An attempt to make clear the position of the individual vertebrae has been made in fig. 3 b, diagrammatic sketch of fig. 3 a. L_5 shows a normal body, separated from the above vertebra by an almost normal intervertebral space; its neural arch presents, however, a

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definite cleft. The bodies corresponding to the other lumbar segments are almost completely fused, with only poor traces of the intervertebral discs.

The upper portion of the lumbar column shows an uncommon deformity resembling a duplication of the spine: at a level most probably corresponding to L_1 there are, in fact, two distinct vertebral bodies widely spread apart. Both the above vertebra and the one beneath are fork-shaped and partially fused with the duplicated L_1 vertebral body, together outlining a large central hole (33 × 24 mm. on the x-ray film). The neural arches of the first four lumbar vertebrae are completely everted (see the dotted line in fig. 3 b). The fourth transverse process on the right is replaced by a large and deformed osseous spur.

In the lateral film (fig. 4) only the fifth lumbar vertebra is well recognizable. The other segments are nearly completely fused and grossly deformed; intervertebral foramina are encroached upon by bone and the posterior arches form a quite irregular fusing osseous mass.

Sacrum and pelvic girdle. No abnormalities are seen, with the exception of asymmetry of the pelvis.

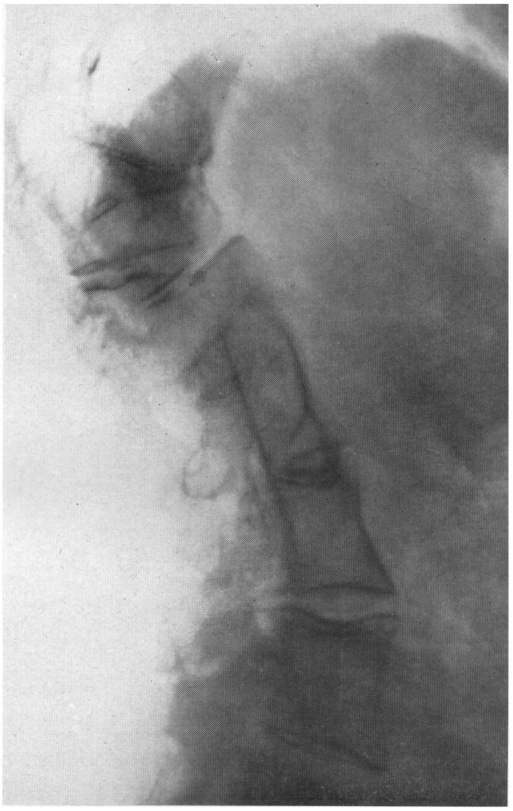

Fig. 4

Discussion

The main deformities in our patient are, briefly: vertebral fusions, multiple clefts of the neural arches, complete gap of the upper lumbar spine, dorsal hemivertebra, abnormal curvatures of the spine, left high scapula, malformations of the ribs. It was not possible to perform a myelography; since, however, such extensive deformities of the vertebral column are known to be usually associated with malformations of the nervous structures (3, 6), the wide cleft of the lumbar vertebrae in our patient is likely to have involved also the spinal cord (diastematomyelia). In addition, an associated protrusion of the meninges in the lumbar region is also probable in our case, on account of the extensive opening of the lumbar neural arches. Such hypothesis is supported by the presence of the lipomatoid protuberance already described in the lumbar region: it is well known, in fact, that true lipomas are often present

n those cases of spina bifida involving the cord and its membranes (« spina bifida with tumor », Leveuf).

With regard to the neurological signs exhibited by our patient, they may be related either to a concomitant involvement of the spinal cord and meninges, or to a direct pressure of the bone on the nerve roots at the level of the deformity.

In the great variety of defects showed by our case, the most interesting deformity is located at the upper lumbar level, where the spine seems to be duplicated. Such an anomaly is extremely rare in the adults and only a few cases have been described in dead fetuses or non-viable infants, usually in association with other lethal abnormalities. A case has been also described in a calf by Adelmann (1).

One of the first cases studied from both radiographic and anatomic point of view, was reported by Lucksch (19), regarding an anencephalic monster with complete separation of the entire spine into two halves and dorsal protrusion of the whole digestive tract.

Feller and Sternberg (6) described two female fetuses showing a complete gap in the cervical and upper dorsal region of the spine.

Other cases have been reported by Korff (16) and by Hartmann (11). The female still-born infant described by Hartmann showed anencephalia, posterior spina bifida throughout the entire vertebral column, and complete separation of the cervical and upper dorsal spine into two distinct halves. On histological examination the author found notochordal remnants on both the two halves of the doubled cervico-dorsal column and therefore he believed that the anomaly was to be related to a duplication of the notochord.

Rosselet (22), Kirchhoff and Rohwedder (13), and Zunin (30) have described cases of complete separation of the lumbo-sacral spine into two symmetric branches in new-born infants who died soon after birth.

A case of pseudoduplication of the spine has also been reported recently by Weigel and Bach (29) in a 53 year old woman, who showed a wide gap of both the bodies and the neural arches from D_7 to D_{12}, associated with other anomalies, namely lumbar butterfly-vertebrae, multiple vertebral blocks, and posterior spina bifida of many other vertebrae.

The pathogenesis of these forms of complete separation of both the vertebral bodies and the corresponding neural arches simulating a duplication of the spine, cannot be fully understood without taking into account the early stages of development of the vertebral column (5, 10, 20, 24).

In the third embryonal week the mesoderm on each side of the closing neural canal becomes segmented into paired lateral masses, the so-called somites. The ventro-medial portion of each somite, termed the sclerotome, breaks down and its cells migrate toward the mid-line, thus joining those from the opposite side. In this way the notochord, which is the very earliest evidence of the axial skeleton, is sorrounded by an axial mass of mesodermal tissue made up of cells originating from the two sides: the ventral portion of this mesenchymal mass will form the vertebral body; the dorsal portion sorrounds the neural canal and will give rise to the neural arches.

At about the end of the fifth week chondrification stage begins with two cartilaginous centers for the vertebral body, separated by a sagittally-placed perichordal septum, and one center for the neural arch on each side (5, 10). According to Schinz and Töndury (24), however, the vertebral body is provided first with two symmetric precartilaginous centers, fusing into one precartilaginous body and soon thereafter followed by a single cartilaginous center. During the cartilaginous stage the notochord becomes constricted by the rapid growth of the cartilaginous vertebra and later entirely disappears. At approximately the tenth week of fetal life ossification begins from three separate centers, one for the vertebral body, and one on each side for the neural arch.

Lack of fusion of the two chondrification centers of the vertebral body, due to persistence either of the perichordal septum or of the notochord, may account for a simple linear cleft in the vertebral body, i.e. for a butterfly-vertebra (5, 10, 12, 15). On the other hand, the genesis of so complex a vertebral malformation as observed in the lumbar spine of our patient, including extensive separation of both the bodies and the neural arches, should be referred to an earlier stage, namely to non-closure of the sclerotomal tissues in the mid-line around the notochord (22). This view is strongly supported also by the fact that in our patient the vertebral split is closely associated with other disorders, such as vertebral fusions, which have been demonstrated (28) to originate in the blastemal stage. On the basis of histologic examinations, some authors (2, 11) believe that these forms of wide rachischisis are due to primitive duplication of the notochord: this is followed by lack of fusion of the sclerotomic cells in the mid-line and development of two separate hemicolumns with one neural hemiarch on each side. This opinion is also shared by other authors (6, 13, 28, 29).

The etiology of the malformations observed in our patient is difficult to be assessed, since only radiologic data are available and suitable investigations on her relatives could not be performed. It is to be noted, however, that very often many of the anomalies seen in our case — namely vertebral fusions, defects in the neural arches, dorsal hemivertebra, scoliosis, high scapula, and rib deformities — show hereditary incidence; therefore it is reasonable to assume that the complex malformative picture of our patient may have likewise a hereditary basis. Since the deformities observed in our case are essentially manifestations of two primary faults, i.e. « schisis » and « synostosis » of the vertebral segments, they may be considered a manifestation of the « schysosynostosis axialis congenita familiaris » (Gedda and Iannaccone, 1956), a hereditofamilial disease of the axial skeleton, which may have many anatomico-clinical expressions: cranio-rachischisis, Klippel-Feil syndrome (8), congenital osseous torticollis (7), block vertebrae, spina bifida.

Since in our patient a hereditary or familial nature of the deformities could not be actually demonstrated, it cannot be excluded that she represents a sporadic case, due to environmental factors acting on the axial skeleton very early during the endo-uterine life.

Another difficult problem concerns the exact terminology for a vertebral anomaly of the type observed at the lumbar level in our patient.

« Spina bifida », although this term is commonly rather loosely used to cover a wide range of defects characterized by fissures of the spine, in its simplest form is merely a defect of the neural arch (5, 10, 15, 20, 23, 25), and therefore is not appropriate to indicate the anomaly in our case. « Somatoschisis » or « spina bifida anterior » are also inadequate, since these words properly define a cleft limited to the vertebral body (15, 18, 21, 22). « Rachischisis » is used with different meanings by various authors. According to some, the term denotes a congenital fissure affecting several neural arches (5, 26), and even the posterior arches of the entire column (14). (« rachischisis totalis »). Other authors call « rachischisis » any defect of closure of the bony spinal canal (3, 4, 9), located either on the anterior wall (« rachischisis anterior »), or on the posterior (« rachischisis posterior »). The term « rachischisis » is, finally, sometimes used to denote a complete sagittal fissure of the whole vertebra, affecting both the body and the neural canal (22, 23, 28).

Rosselet (22), describing a case of complete gap of the lumbo-sacral spine, claims that the most suitable terms for anomalies of this type would be « rachischisis antéro-postérieur total » or « colonne dédoublée ». It does not seem justified, however, to speak of a true duplicated spine, since in these deformities there is only a half neural arch on each side. On the basis of the above recalled embryologic data it should be clear that the vertebral malformations of this type represent a true « schisis », a term which properly denotes a cleft resulting from lack of fusion of two parts which originate from two distinct "anlagen,, and normally should fuse together. Therefore, the most suitable term for the lumbar deformity presented by our patient and the few other cases described in the literature, seems to be « rachischisis » (etymologically: cleft of the spine), in the sense of a complete separation of the whole vertebrae in the sagittal plane.

Summary

A case is described of uncommon vertebral malformation simulating a duplication of the spine. Pathogenesis, etiology and terminology of such an anomaly are briefly discussed.

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RIASSUNTO

L'autore descrive una rara malformazione vertebrale, che simula uno sdoppiamento della colonna in sede lombare; discute inoltre brevemente la etiopatogenesi e la esatta terminologia di tale deformità.

RÉSUMÉ

L'auteur décrit une rare malformation vertébrale réalisant l'image d'une colonne dédoublée et discute brièvement la pathogénie, l'étiologie et la terminologie de cette malformation.

ZUSAMMENFASSUNG

Der Verfasser beschreibet eine seltene Fehlbildung, die eine doppelte Wirbelsäule scheint, und bespricht Kurz die Pathogenese, die Aetiologie und die terminologie derselben.

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