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BILATERAL SPRENGEL'S SYNDROME WITH SITUS INVERSUS TOTALIS

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The main abnormality in the syndrome that carries the name of Sprengel (1891) is a congenital abnormally high location of the shoulder blade. The condition occurs equally in males and females and both sides are evenly involved. In only 10 per cent of cases is the deformity bilateral.

In this syndrome the shoulder blade itself is hypoplastic and its cranial part, containing the supraspinate fossa, is elongated and protrudes ventrally. The scapula is rotated so that the spina scapulae runs upwards towards the midline, whereas the distal angulus scapulae points away from the thoracic cage. This results in a slight endorotation of the upper arm and some loss of abduction, flexion, and extension in the shoulder joint. The loss of function may be aggravated by shortening and contracture of the shoulder muscles. Many of these patients therefore prefer to wear the arm on their back. The high location of the shoulder forces them to keep the head forward and slightly rotated towards the normal side, which may cause the false impression of a torticollis (Ombrédanne 1944).

Concomitant malformations of the cervicothoracic spine and of the chest are frequent (Aschner & Engelmann 1928, Jeannopoulos 1952, 1961). In addition to this, about one-third of all patients show a very typical abnormity: a fibrous, chondro-ossal or bony bridge between the cervical spine and the scapula. When it is made of bone it is called an *os omovertebrale*. It lies between the spinous and transverse processus of the lower cervical vertebrae and the medial and caudal margin of the scapula. At both ends it may be fixated by fibrous tissue or by bone fusion and sometimes the connection contains a real joint or a pseudarthrosis.

In many cases there is a gradual transition from Sprengel's deform-

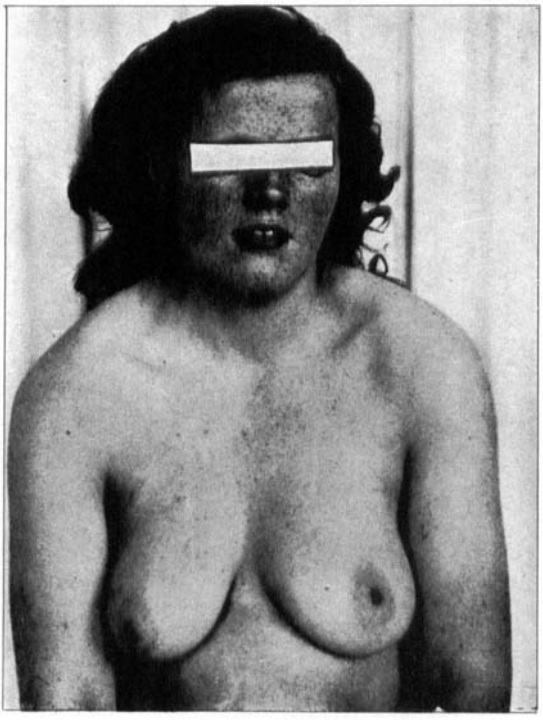


Figure 1. The patient shows lentigo pigmentations and a remarkable contour of the shoulders. The throat is normal.

ity to other congenital malformations at the cervicothoracic level (Schrock 1949, Matzen & Fleiszner 1970) such as the syndrome of Klippel & Feil (1912). The two may even combine with platybasia of the skull, presenting the syndrome of Furst & Ostrum (1942). These combinations of malformation and the transitions of one syndrome into another make it probable that some exogenic embryopathy c.q. myelopathy underlies all these dysraphic conditions (Bijl 1956, Degenhardt 1964). Information about the familiar character of Sprengel's syndrome is scarce and most descriptions concern solitary cases, but Gottesleben (1927) reported about a family in which the syndrome was present in three generations and Schwarzweller (1937) saw nine families in which the syndrome itself or the concomitant deformities of the chest and the spine were frequent in several generations. Therefore, the next case report may be of interest and even more so since it presents a hitherto unknown combination of syndromes.

In 1948 we observed a young woman, born in 1926, for vague upper abdominal complaints. She showed extensive lentigo pigmentation, mainly in the face and a remarkable posture (Figures 1 and 2). Both

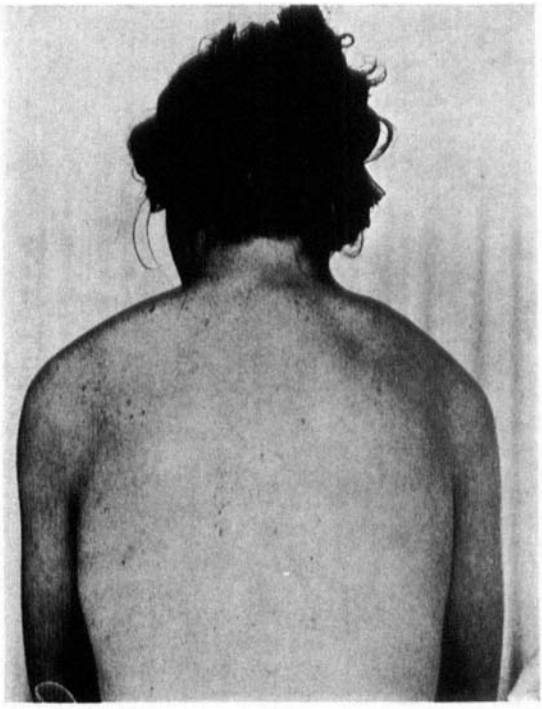


Figure 2. Although the shoulders run up very high, there is still a well-defined neck.

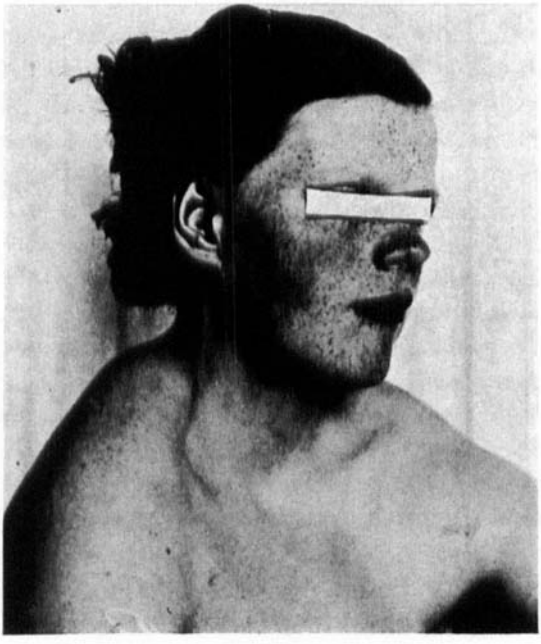


Figure 3. The fossae supraclaviculares are exceptionally deep.

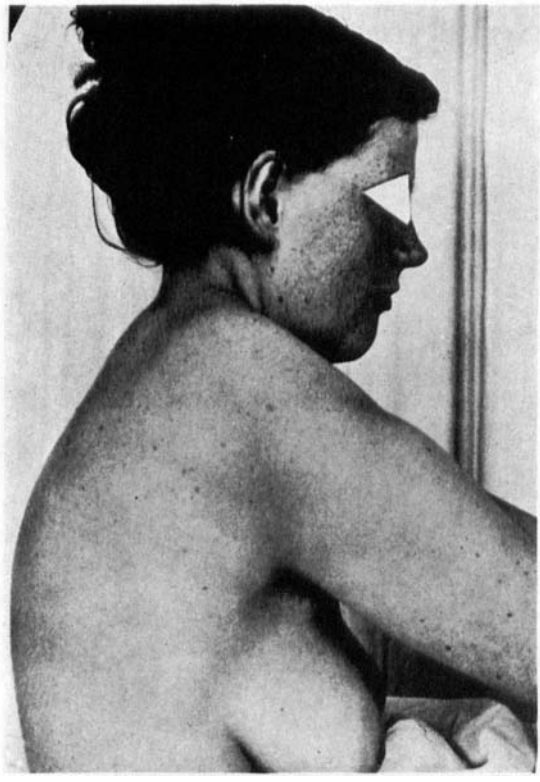
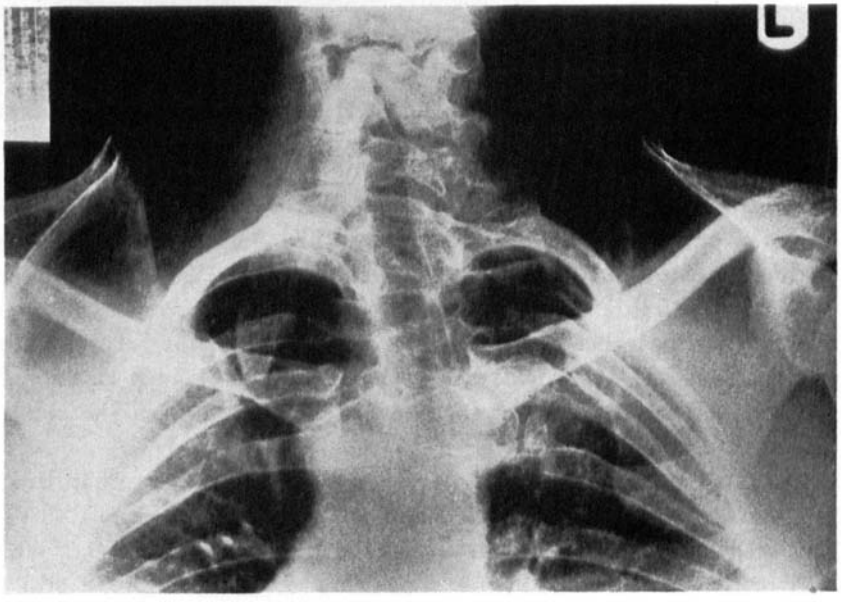


Figure 4. There is some thoracic kyphosis.

Figure 5. Typical rotation and deformation of the scapulae; os omovertebrale on the right.



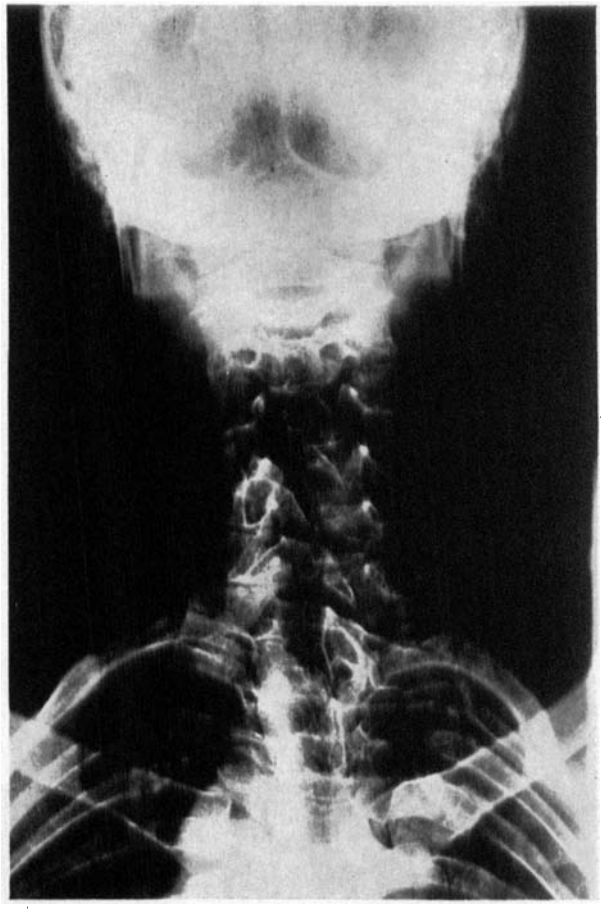


Figure 6. Fission of vertebrae; a large cervical rib on the right side and a small one on the left.

shoulders stood very high, especially on the right side, with deep and large supraclavicular fossae (Figure 3). She had a slight thoracic kyphosis (Figure 4). The neck and the throat were well defined, rotation and flexion of the head were normal, but extension was limited to 30°. Both shoulder joints showed reduced abduction, elevation and exorotation, but the patient did not consider herself incapacitated in any way. In fact she performed rather heavy work in a grocery store. Between the scapula and the lower cervical spine on the right side a bridge of hard tissue could be felt.

The X-ray examination shows a typical os omovertebrale in this area (Figure 5) between the caudal angulus of the scapula and CV, VI and VII without bony fusion and connected to these structures through a real or pseudo joint. The high position of both scapulae and their

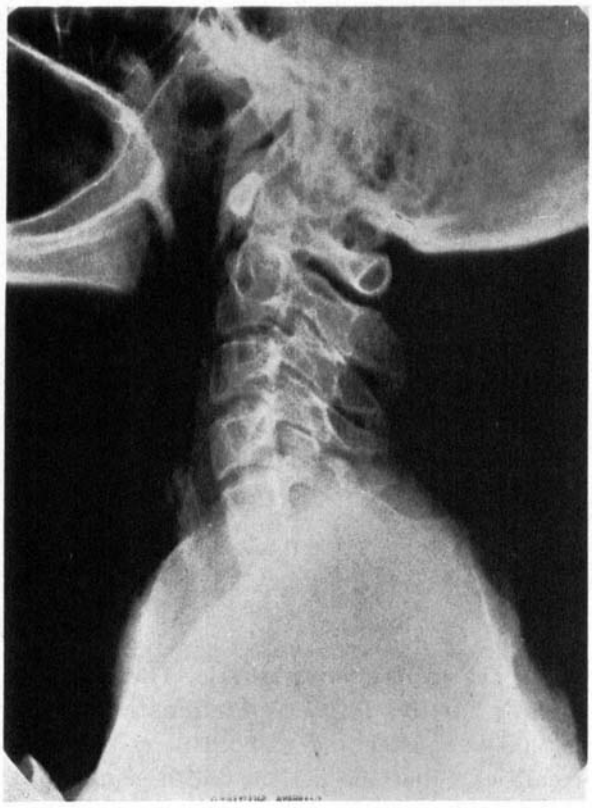


Figure 7. Fusion of CV and CVI. The X-ray is made in maximum lordosis of the cervical spine.

deformation is apparent. They are rotated so that the spina scapulae runs up towards the midline. The clavicles point far upwards in lateral direction. There is a large cervical rib on the right and a small one on the left and the bodies of CV, VII, Th I and II are split (Figure 6). The cervical lordosis is slight, as shown in Figure 7, made in maximum extension. Figure 7 also shows fusion between CV and VI. The dorsal curves of the first 5 thoracic ribs were flattened out, especially on the left, and there was a fusion between costa I and II on the left. The skull and the rest of the skeleton were completely normal. The chest films (Figure 8) confirmed the clinical diagnosis of dextrocardia: the heart lies in the right hemithorax. Further X-ray examination made it clear that the stomach was on the right side of the abdomen and the gall bladder was on the left, as was the coecum.

This then is an exceptional case of bilateral Sprengel's deformity with situs inversus totalis and with several concomitant deformations

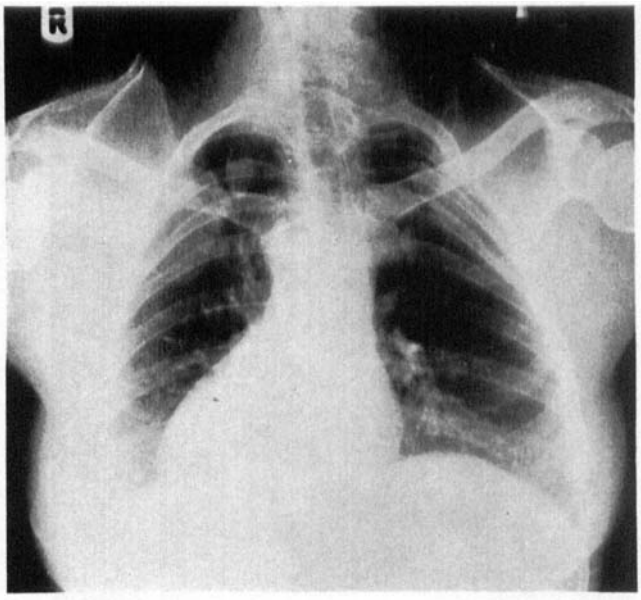


Figure 8. Dextrocardia, combined with situs inversus abdominalis.

in the cervicothoracic region, *i.a.* an os omovertebrale. The condition was unknown in the patient's family. Physical examination of her parents, her two brothers, and one sister and of both pairs of grandparents showed no abnormalities. Since the patient had no complaints from her deformity and further observation did not show any abdominal pathology that needed treatment, she was discharged from the hospital.

We saw her again recently. She has been in good health since 1948 and has married a normally built husband. They have 5 children, three boys and two girls. The youngest boy and the oldest girl are twins. None of them has any clinical symptom of Sprengel's deformity or situs inversus. The X-rays of the family are presented in Figure 9. They were all made with maximum depression of the shoulder. None of the children shows any abnormal configuration of vertebrae or ribs and they have no dextrocardia. The scapulae are not deformed and do not lie in an abnormally high position. However, in the children I, II, and IV there is some rotation of the scapula, especially on the right side. It causes an abnormal direction of the spina scapulae and elevation of the medial upper corner of the shoulder blade. Comparing the pictures with those of the father or of the other children makes this very clear indeed.

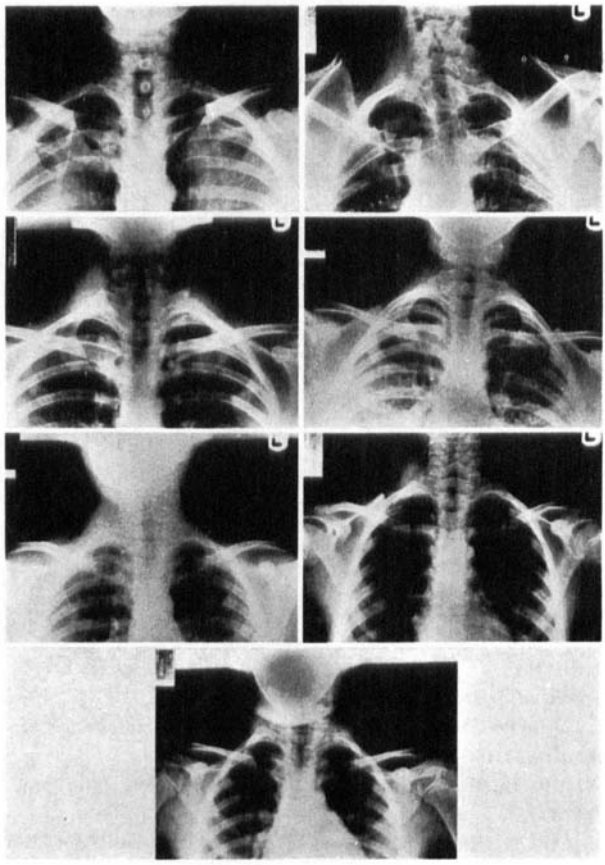


Figure 9. Shoulder girdles of the family.
At the top, the normal husband and his wife with Sprengel's deformity and situs inversus totalis. I and II: Two boys with slight rotation of the scapulae. III: A boy with a perfectly normal position of the scapulae. IV: His twin sister shows rotating scapulae, especially on the right. V: A girl without any malformation.

All children show a density in the soft tissues on the right side of the neck. It does not correspond to any palpable finding and on the original pictures it is less evident. Therefore we are not quite sure whether we may really interpret it as a remainder of the mother's deformity, which is undoubtedly true for the rotation that some of the children still show.

This family, then, is a good example of the way a pronounced Sprengel's deformity may appear suddenly and again disappear in the offspring, leaving in them only slight traces of the original malformation.

SUMMARY

The author describes a female patient with bilateral Sprengel's deformity combined with several congenital malformations in the cervicothoracic region, *inter alia* an os omovertebrale on the right side. Of more interest and more exceptional is the fact that the patient also has a complete situs inversus. The deformity does not occur in her grandparents, her parents, or her brothers and sister.

She married a normal husband and has 5 children. None of these has Sprengel's syndrome or situs inversus, but in three of the children the scapulae show a slight rotation.

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