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CONGENITAL SYNOSTOSIS IN THE CERVICAL SPINE

By

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The first detailed account of this syndrome was given in 1912 by *Klippel & Feil* from Paris. Until 1940 a total of 133 cases had been published.

Among several signs in the pronounced cases of the syndrome, the most important are the following: the short neck with very restricted movements, the so-called wing-neck or pterygium colli and the low hairline at the back of the head.

The purpose of this paper has been to determine the frequency of the syndrome in the population of Copenhagen, and the nature and frequency of the accompanying deformities. The material comprises 76 cases collected from the archives of all X-ray departments in Copenhagen. In the years 1951–60 25 new cases were diagnosed. An estimation based on these figures results in a total of approximately 200 cases in Copenhagen, *i.e.* 0.2 per thousand. Probably this is a minimum figure, because several mild cases would not be diagnosed on account of the lack of clinical symptoms. In some of the patients the synostoses were found quite accidentally by an X-ray examination of the trachea.

Table 1 shows the localization of the synostoses in the cervical spine. A classification is made according to the number of synostoses: 44 patients with one synostosis (between the vertebral bodies), 19 patients with two to five synostoses and 13 patients in whom all the vertebral bodies in the cervical spine are fused together. However, it is to be noted that the atlas is not involved in six of the 13 pronounced cases and not in any of the milder cases.

In the mild cases the synostoses seldom continue down in the thoracic spine, in the pronounced cases nearly always (Table 1), even to the eighth thoracic vertebra. The frequency of the synostoses is highest in

the upper part of the cervical spine and decreases gradually downwards. It is a curious fact that a single synostosis between the third and fourth body seldom occurs, but in the 19 cases with two to five synostoses this localization is the most frequent.

TABLE 1

Localization	Synostosis: Number		
	One 44 cases	Two-five 19 cases	Six or more 13 cases
1.–2. cerv. vert.	0	0	7
2.–3. „	13	13	13
3.–4. „	3	16	13
4.–5. „	7	13	13
5.–6. „	7	8	13
6.–7. „	4	7	13
7. cerv. and 1. dorsal vert.	1	2	11

TABLE 2

Other deformities	Number of synostoses			Total 76 cases
	One 44 cases	Two-five 19 cases	Six or more 13 cases	
In the cervical spine	57 %	47 %	77 %	58 %
In the rest of the spine	54 %	44 %	100 %	62 %
Scoliosis	...	...	...	71 %
Scoliosis in the cervical spine	31 %	47 %	38 %	36 %
Scoliosis in the rest of the spine	46 %	56 %	23 %	44 %
Cervical rib	11 %	11 %	31 %	14 %
Sprengel's deformity	9 %	11 %	31 %	16 %

Table 2 shows the frequency of the accompanying vertebral deformities, expressed in percentages. The subdivision is the same as in Table 1. In the column to the right the entire material is presented.

In the cervical spine, accompanying deformities—usually clefts in the arches—are found in roughly half of the cases, most frequently in the cases with six or more synostoses. In the rest of the spine, other deformities—frequently synostoses—are seen in about half of the mild cases but in all the pronounced cases, pointing to the fact that the cervical

spine is not the only localization of the syndrome, but should be considered as the topographical center of the syndrome—frequently the skull is involved too.

Scoliosis is found in 71 per cent of the cases, apparently more frequently in the mild cases and outside the cervical spine. Cervical ribs and Sprengel's deformity are seen in 1/10 of the mild and 1/3 of the pronounced cases respectively.

The cause of the deformities is unknown, but apparently the disease might be hereditary. For obvious reasons comparison could be made with Turner's syndrome; patients with this syndrome sometimes are shortnecked and wingnecked just as patients with Klippel-Feil syndrome sometimes show hormonal insufficiency.

Finally it should be mentioned that recently it has been proved that X-ray irradiation of pregnant mice results in various deformities in the foetuses especially in the cervical spine.

SUMMARY

The material comprises 76 cases and it is estimated that the syndrome is found in 0.2 per thousand in the population of Copenhagen. The synostoses are most frequently found in the upper part of the spine, in 13 cases the cervical spine is completely synostosed. Other vertebral deformities in the whole spine are found in about 50 per cent, scoliosis in 71 per cent. Sprengel's deformity in 16 per cent of the cases.

RESUME

Le matériel d'observation compte 76 cas et on calcule que le syndrome existe chez 0,2 p. mille de la population de Copenhague. Les synostoses sont observées le plus fréquemment dans la partie supérieure de la colonne vertébrale; dans 13 cas, la colonne cervicale était complèment synostosée. D'autres déformités vertébrales dans toute la colonne ont été trouvées dans environ 50 % des cas, la scoliose dans 71 %, la déformité de Sprengel dans 16 % des cas.

ZUSAMMENFASSUNG

Das Material umfasst 76 Fälle und man schätzt, dass das Syndrom bei 0,2 per Tausend in der Bevölkerung von Kopenhagen zu finden ist. Die Synostosen sind am häufigsten im oberen Teile der Wirbelsäule zu

finden. In 13 Fällen war die Halswirbelsäule vollständig synostosiert. Andere Missbildungen in der ganzen Wirbelsäule wurden in ungefähr 50 % gefunden, Skoliose in 71 %, Sprengels Missbildung in 16 % der Fälle.

REFERENCE

Klippel, M. & Feil, A.: Un cas d'abscence des vertebres cervicales. Nouv. Icon. Salp., 25: 223–250, 1912.