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VERTEBRAL MALFORMATIONS AND ASSOCIATED SOMATICOVISCERAL ABNORMALITIES

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Numerous references to spine abnormalities and associated somatic and/or visceral abnormalities are reported. An implication is present in each that the association is more than just coincidental and some cause and effect relationship might exist. A series of cases was collected in which congenital spine abnormalities were noted on roentgenograms from patients evaluated for varied reasons. Other roentgenograms of these patients and their medical records were reviewed in an attempt to find associated congenital abnormalities. Another group of patients with known congenital visceral malformations had their roentgenograms reviewed to see if they had spinal variations. It was found that patients with congenital vertebral malformations had an extremely high rate of associated visceral abnormalities, especially of the heart and kidneys. Other relationships between the spine and viscera include congenital lesions of the gastrointestinal and respiratory system and other parts of the renal system. This study also suggests the possible relationship between vesicoureteral reflux and spina bifida occulta.

Embryologic considerations yield likely reasons for the relationships which have been suggested previously and are emphasised and augmented by this investigation. The supporting argument that a definite relationship exists between spinal variations and visceral abnormalities, as well as the embryologic reasons, is presented in this review of our selected cases and the appropriate literature.

An association between congenital vertebral abnormalities and other somatic and visceral abnormalities has been referred to by many authors (Arcomano and Azzoni, 1967; Elliot *et al.*, 1970; Lynch and Grisson, 1966; Rosenfelder, 1968). Theories have been postulated to explain the reason for the relationship, and the findings of some pathological material does support the logic of the theories.

The author was intrigued by the frequency with which somatic and visceral abnormalities were found in association with common and less common spine variations. Because of that interest, an attempt to answer questions about spinal dysrhaphism in a large group of cases was undertaken. The questions of primary concern were: (1) What is the frequency with which somatic and visceral abnormalities occur with spinal dysrhaphism? (2) Is the level of the spinal abnormality related to the presence of a specific malformation? (3) Are the vertebrae with specific anatomical variations found more often with specific somaticovisceral anomalies? and, (4) With what frequency do patients with extra-axial somatic or visceral abnormalities have spinal abnormalities?

An analysis of our material and review of the pertinent literature was undertaken to answer those questions.

MATERIALS AND METHODS

All cases with variations of the spine recognised during the routine of daily film interpretation were recorded. This procedure was followed for approximately two years (1970 and 1971). The available records of those patients were then analysed to see what associated abnormalities were documented. The patients were not subjected to a routine or standardised investigation.

Roentgenograms of patients with known 'congenital' visceral abnormalities were reviewed as were their medical records. The roentgenograms of these patients were selected at random from the congenital cardiac and congenital genitourinary tract files. film The number of patients with and

TABLE I

Type of abnormality	*Number of patients*	*Somaticovisceral abnormality*
Dorsal vertebral abnormalities		
Spina bifida occulta	(1)	Transposition and PDA (1)
Fusion	(3)	Large kidneys (cause?) and reflux (1)
		Oesophageal atresia (1)
		VSD pulm. st. (1)
Block vertebrae	(1)	Tetralogy (normal IVP) (1)
Hemivertebrae	(2)	Mitral atresia, single ventrical and hypoplastic aortic arch (1)
		VSD (1)
Asymmetrical butterfly vertebrae with fusion	(3)	Isolated dextrocardia (1)
		Shunting lesion (?VSD) (1)
		Tetralogy (1)
Symmetrical butterfly	(1)	Tricuspid stenosis, VSD; PDA (1)
Hemivertebra and asymmetric butterfly	(1)	VSD, Rt. Arch, coarctation (1) aortic
Lumbar vertebral abnormalities		
Spina bifida occulta	(17)	Vesicoureteral reflux (10)
		UPJ obstr. and post. values (1)
		UVJ obstr. (1)
		UPJ obstr. (1)
		Tetralogy and solitary kidney (1)
		No abnormality (3)
Fusion	(1)	Imperforate anus and solitary kidney (1)
Spina bifida	(3)	Symmetrical dysrhaphism with Arnold–Chiari II malformation; normal GU tract (3)
Sacral vertebral abnormalities		
Spina bifida	(1)	Meningomyelocoele (1)
Spina bifida occulta	(6)	No abnormality (3)
		Vesicoureteral reflux (1)
		Hydrometrocolpos (1)
		Solitary kidney (1)
Partial absence, fusion and curved sacrum	(4)	Sacral meningomyelocoele (1)
		Renal at., hydrometrocolpos (1)
		Double vagina, UVJ and renal dupl. (1)
		Ambiguous genit., exstrophy, cloacal abnormality, bifid vagina, septate bladder (1)
Hypoplasia	(1)	VSD, imperforate anus, TEF (1)
Lumbosacral vertebral abnormalities		
Spina bifida occulta	(8)	Vesicoureteral reflux (2)
		Sacral dermal sinus, and ? lipoma (1)
		UPJ obstruction (1)
		Ureteral stenosis (1)
		Bladder diverticulum (1)
		No abnormalities (2)
Symmetrical spina bifida	(6)	Meningomyelocoele and Arnold–Chiari II malformation (6)
Hypoplasia and fusion	(2)	Solitary kidney and imperforate anus (1)
		Renal duplication and ectopic ureteral orifices (1)
Aplasia and fusion	(1)	Imperforate anus, malrotation of kidneys (1)
Increased interpedicular distance	(3)	Meningomyelocoele with renal malrotation (1)
		Bladder exstrophy, superior pole renal duplication, ureterocoele (1)
		Negative (1)
Severe gibbus	(1)	Negative (1)

TABLE 1 – *continued*

Type of abnormality	*Number of patients*	*Somaticovisceral abnormality*
Dorsal-lumbar vertebral abnormalities		
Asymmetrical butterfly, hemivertebrae, fusion	(1)	Solitary kidney, reflux (1)
Asymmetrical butterfly, hemivertebrae	(1)	Hydrometrocolpos, CHD (specific?) (1)
Thoracic spina bifida, bar, scoliosis, fusion	(1)	Meningomyelocoele, normal GU tract (1)
Symmetrical butterfly (dorsal) and asymmetrical butterfly (lumbar)	(1)	Absent left kidney and ectopic perineal anus (1)
Asymmetrical butterfly with fusion	(1)	No abnormality, ? CHD (1)
Symmetrical butterfly – dorsal and lumbar spina bifida	(1)	Diastematomyelia and hydrocephalus (1)
Asymmetrical butterfly with fusion and spina bifida	(1)	Meningomyelocoele, otherwise normal (1)
Symmetrical fusion-block	(1)	Normal (1)
Asymmetrical butterfly and spina bifida	(1)	Truncus arteriosus with ventricular inversion, meningomyelocoele (1)
Sacrococcygeal vertebral abnormalities		
Curved (sickle shape) only	(1)	Short urethra, renal dysplasia (1)
Curved sacrum, absent coccyx	(1)	Absent left kidney, double vagina, rectovesical fistula (1)
Sacrococcygeal hypoplasia	(1)	Imperforate anus, rectal fistula (1)
Spina bifida occulta, curved and foreshortened	(1)	Exstrophy of bladder (1)
Thoraco-lumbar-sacral		
Asymmetrical butterfly (T-1), aplasia of lumbar vertebrae caudad	(1)	Single 'cake' kidney (1)
Spina bifida, asymmetrical with fusion	(1)	Meningomyelicoele with hydrocephalus (1)
Spina bifida, symmetrical	(4)	Meningomyelocoele malrotation kidneys (1) Meningomyelocoele 'kissing' ureters (2) Meningomyelocoele only (1)
Hemivertebra, butterfly with fusion, absent sacrum	(1)	Imperforate anus, rectovesical fistula, crossed renal ectopy (1)
Multisegment with fusion and partial sacral absence	(1)	Malposition kidneys, ? CHD (1)
Cervico-dorsal-lumbo-sacral		
Multiple hemivertebrae	(1)	Imperforate anus, absent left thumb (1)
Thoracosacral		
Butterfly, fusion	(1)	Crossed renal ectopy and solitary kidney (1)
Cervical-thoracic		
Spina bifida occulta	(1)	ASD, VSD (1)
Lumbo-sac-coccygeal		
Spina bifida occulta	(1)	Reflux (1)

TABLE 2

Cardiovascular abnormality	*Spine abnormality*	*Patients with no abnormality*
Heart	—	3
Double aortic arch with vasc. ring (3)	—	8
Arterial septal defect (8)	—	8
Ventricular septal defect (8)	Dorsal butterfly (1)	3
Ductus arteriosus (4)	Spina bifida occulta C6 and 7, T1–7 (1)	
A–V communis (1)	Spina bifida occulta T1–8 (1)	
Anomal. left coronary (1)	—	
Anomal. pulmonary venous drainage (4)	—	3
Total (3)	—	1
Partial (1)	Spina bifida occulta T2–7 (1)	5
Valvular aortic stenosis (6)	Spina bifida occulta T1–7 (1)	5
Coarctation (8)	Spina bifida occulta T1–8 (1)	
	Spina bifida occulta T4–8 (1)	
	Spina bifida occulta T1–9 (1)	
Tricuspid atresia (3)	Fused dorsal vertebrae (1)	1
	Spina bifida occulta T3–7 (1)	11
Pulmonary atresia and stenosis (13)	Spina bifida occulta T3–4, 5 (1)	
	Spina bifida occulta T1–8 (1)	
Pseudotruncus (1)	13 dorsal vert. (1)	10
Tetralogy (12)	Fused dorsal (1)	
	Spina bifida occulta T1, 2, 3 (1)	
Anomal. left pulm. art. (1)	Spina bifida occulta T1–8 (1)	
Truncus arteriosus (1)		1
Double outlet right vent. (1)	Spina bifida occulta T5, 6, 7 (1)	7
Transposition (11)	Spina bifida occulta C7–T6 (1)	
	Spina bifida occulta T1–8 (1)	
	Hemivert., scoliosis (1)	
Corrected transposition (1)		1
Nature of abnormality	*Spine abnormality*	*Number of patients*
Kidney		
Duplication	None	1
Polycystic, infantile	None	4
Nephronopthisis	None	1
Multicystic	None	2
Calyceal diverticulum	None	2
Malrotation	Spina bifida occulta S-1	2
Ectopic kidney, pelvic and solitary	Fusion, L-2 and 3	1
Crossed ectopy with fusion	Block, S-1 and S-2 coccygeal fusion	1
Ectopic kidney (pelvic)	Spina bifida occulta S-1, 2, 3, (high grade)	1
Crossed ectopy – duplication of ureter	Fusion of L-3 and 4, sacral block, constriction abnormality	1
Crossed ectopy – no fusion	Spina bifida occulta L-5, S-1, S-2	1
Crossed ectopy	Elevated arch, L-4, spina bifida occulta L-5, S-1 (high grade)	1
Ectopic kidneys (pelvic)	Decreased interpediculate distance	1
Crossed ectopy	Dorsal butterfly, symmetrical and asymmetrical fusion ? congenital heart lesion	1
Ureter		
Ureterocoele	Elevated arch – L5, and spina bifida occulta S1 and 2	1
Ureterocoele	None	3
Ectopic ureter–vagina	None	1
Ectopic ureter–urethra	None	1
Duplicated ureters	None	1
Ectopic ureter, unspecified	None	3
UPJ	None	4
UPJ	Spina bifida occulta (S-1, S-2), high arch	1
Ectopic ureter with ureteral duplication	None	1

TABLE 2 – *continued*

Nature of abnormality	*Spine abnormality*	*Number of patients*
Bladder		
Duplication (with ureterocoele)	None	1
Duplication	None	1
Urethra		
Urethral divertic. (or embryonic remnant)	Hemivertebra	1
Urethral diverticulum	None	1
Other		
Abnormal renal vasculature	None	3
Urachal cyst	None	1
Urachal cyst	Spina bifida occulta L-5	1

without spinal variations and the nature of those variations were documented. The results of these studies appear below (Tables 1 and 2).

RESULTS

All findings are tabulated in Tables 1 and 2. Comments referable to those data and points worthy of emphasis are included in this section. Of the dorsal vertebral abnormalities, 10 of 12 patients had significant congenital heart lesions. The spina bifida occulta patient had vertebrae D-2, D-3 and D-4 involved with arch diastasis of at least 3 mm. The heart abnormalities involved the left heart twice and the right heart five times; there were four shunting lesions.

In the 'pure' lumbar group of dysrhaphic spine problems 10 of the 17 patients with spina bifida occulta had vesicoureteral reflux. The occult lesion was at L-5 in all instances. Solitary kidneys were noted in two instances; one patient had an L-5 spina bifida occulta and one a fusion dysrhaphism of the lumbar spine. The lumbar abnormalities, other than spina bifida, commonly have associated genitourinary tract abnormalities (13 of 16 patients). The spina bifida patients all had anatomically normal genitourinary tracts.

Spina bifida occulta in the sacral group always involved at least vertebra S-1. S-2 and S-3 were frequently involved (40% of the time). The dysrhaphism (vertebral arch cleft) was at least 4 mm in width. No somaticovisceral abnormality was found in three patients with spina bifida occulta. Fifty per cent of patients in the sacral group had problems related to cloacal differentiation (bladder, rectum or vagina) and 50% had problems directly related to the genitourinary tract.

In the group of patients with lumbosacral vertebral dysrhaphism (21 patients) two of eight with spina bifida occulta had vesicoureteral reflux and two were normal. A lipoma or possibly a dermoid cyst was present in the one who had a sacral dermal sinus. The three others had genitourinary problems. Therefore, five of eight had problems referable to the genitourinary tract. Three patients of 21 had some vertebral fusion abnormality; each had a problem directly related to the kidney.

Nine patients had dorsal-lumbar spinal abnormalities and two of these had congenital heart abnormalities; one other was suspected of having a congenital heart lesion. Three patients had genitourinary anomalies. Four patients had spina bifida; all of these had associated congenital vertebral anomalies. One of the four had a spina bifida with a symmetrical lesion (butterfly vertebra), a lesion of the spine (diastematomyelia), and the often associated hydrocephalus. Of the three 'asymmetrical' spina bifida patients all had meningomyeloceles and one had congenital heart disease. Four patients had sacrococcygeal abnormalities. All had lesions related to pelvic structures and two had renal anomalies.

Eight patients had lesions involving a portion of the dorsal (thoracic), lumbar and sacral vertebrae and/or arches. One may have had a congenital heart lesion (unproven). Five had spina bifida malformations. None of the five had any significant associated somaticovisceral abnormality; the kidneys were malrotated slightly in one and the ureters were close together in their middle third portions in two. Fusion problems were evident in three patients. One of these had spina bifida with no genitourinary tract abnormality. Three patients with aplasia and/or fusion abnormalities had either renal fusion ('cake' kidney) or significant displacement of the kidneys.

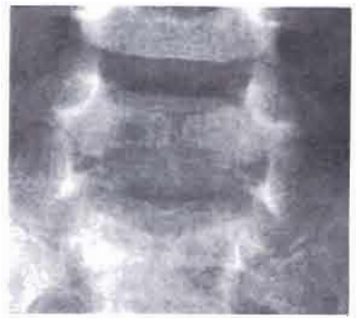

FIG. 1
A spina bifida occulta at L-5. Separation of the two primary elements of the arch is approximately 2 mm (on the original X-ray).

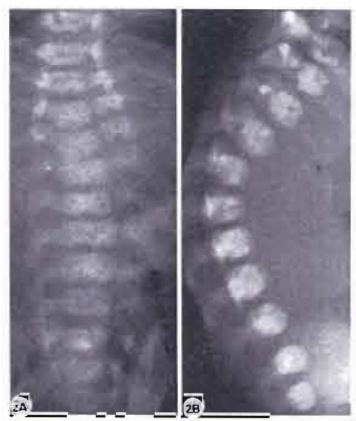

FIG. 2
A. A symmetrical spina bifida (cystica). Note the vertebral bodies are all present, well aligned, and if deformed are symmetrical. The associated arch abnormalities are also symmetrical.
B. Lateral view of the spine of the same patient. A meningomyelocele is associated.

Of the remaining four groups, there was only one patient as a representative per 'group'. The patient with a fusion abnormality in the sacrum (the 'thoracosacral' patient) had a solitary kidney which was ectopic. The patient in the 'cervical-thoracic' group had a large spina bifida occulta and heart disease and the one patient with lumbosacral-coccygeal dysrhaphism had reflux. A patient with numerous hemivertebrae (cervico-dorsal-lumbo-sacral patient) had multiple congenital anomalies including an imperforate anus and absent thumb.

Table 2 shows the results of the spine evaluation in patients with known congenital heart lesions. Ninety-one patients are in the group. Four show unequivocal significant dysrhaphism (4·4%). The dysrhaphism in this report includes spina bifida occulta. Assuming spina bifida occulta is referable to a discussion of dysrhaphism, then patients with congenital heart lesions have associated dysrhaphism 21% of the time (19 of 91 patients). Table 2 also documents the results of an evaluation of 53 patients with known anomalies of the kidney, ureters and bladder. In those patients with renal abnormalities, the most frequent vertebral abnormality was a fusion or block malformation. Patients who have displaced, malrotated, absent, or fused kidneys have a much higher incidence of vertebral malformations. The incidence in this series approaches 60% even if patients with spina bifida occulta are not considered. Some problems related to the kidney (dysplasias), ureters (ectopy, duplications, ureteroceles, congenital obstructions), bladder (duplication) and urethra (diverticulum) were infrequently associated with vertebral abnormalities in this series.

DISCUSSION

Nomenclature. – The term spinal dysrhaphism is used here in its most general sense, that is, any deviation of the vertebral body or neural arch from a normal growth pattern is considered spinal dysrhaphism (Burrows, 1968; Dharia, 1969). The arch is included because it has been shown that some of the factors influencing its development are integrally related to the factors involved with the development of the vertebral bodies and all variations of the growth of the body and arch should be considered related (Källen, 1968). Spina bifida occulta (Fig. 1) refers to a cleft in the arch of a vertebral body usually greater than 1 mm in width. In some instances the clefts were greater than 1 mm in width, however, moderate or gross arch dysplasia

was not considered simply spina bifida occulta in this series. A more severe form of spina bifida occulta is true spina bifida. This term is used to define a significant dysplasia, or even aplasia of the arch, almost universally associated with a meningocele or meningomyelocele (Barson, 1970; Brocklehurst, 1971) (Fig. 2A, B). The interpediculate distance is almost always widened and the pedicles and/or vertebral bodies are also of increased width and/or malformed. Spina bifida may be associated with normal appearing vertebral bodies, moderately deformed vertebral bodies (with or without bilateral symmetrical malformation), or severe vertebral body malformation (i.e. associated with any of the malformations noted below) (Fig. 3).

Fusion abnormalities of vertebral bodies is used here to indicate any congenital conjoining of usually separated portions of vertebral bodies even if those portions were significantly deformed (Fig. 4). It does not attempt to distinguish between fusions of hypersegmentation or hyposegmentation origin.

Block vertebrae are called such when all landmarks of the vertebral masses involved are distinguishable and well maintained with the exception

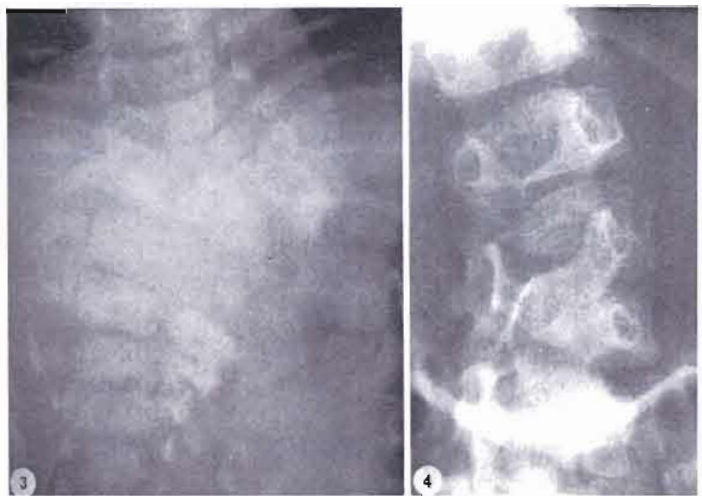

FIG. 3
Asymmetrical spina bifida. The vertebral bodies are malformed in an asymmetrical fashion, associated with hemivertebrae, other abnormal segmentation anomalies and fusions. Associated arch anomalies are also severe and dissimilar from side to side. A meningomyelocele is associated.

FIG. 4
Fusion anomalies involving L-2 and L-3. The anomaly here may represent hyposegmentation or hypersegmentation (see text).

of the joint spaces between them (which were never formed) (Fig. 5A, B). Butterfly vertebrae are those with a central constriction and flared lateral margins (or relatively heightened lateral margins) Colquhoun, 1968). Obviously the shape on the anteroposterior view of a spine roentgenogram is that of a butterfly with wings spread (Fig. 6). 'Symmetrical' and 'asymmetrical' were terms used relative to the butterfly vertebrae and distinguish those vertebrae with bilateral symmetrical deformities from those in which some or considerable asymmetry was obvious. A hemivertebra is a butterfly vertebra with only one wing (Fig. 7). Again, this vertebral unit may be the result of hypersegmentation (too many elements formed – the hemivertebra itself) or hyposegmentation (not enough elements formed – the remaining portion). The other terms used to describe the abnormalities herein discussed are self-explanatory and include aplasia, hypoplasia, curved (i.e. sickle shaped), foreshortened (i.e. hypoplasia at the end of a structure – such as sacrococcygeal foreshortening) and combinations of some of the terms noted above.

Embryology. – After fertilisation of the egg cell, replication occurs and differential cell movements

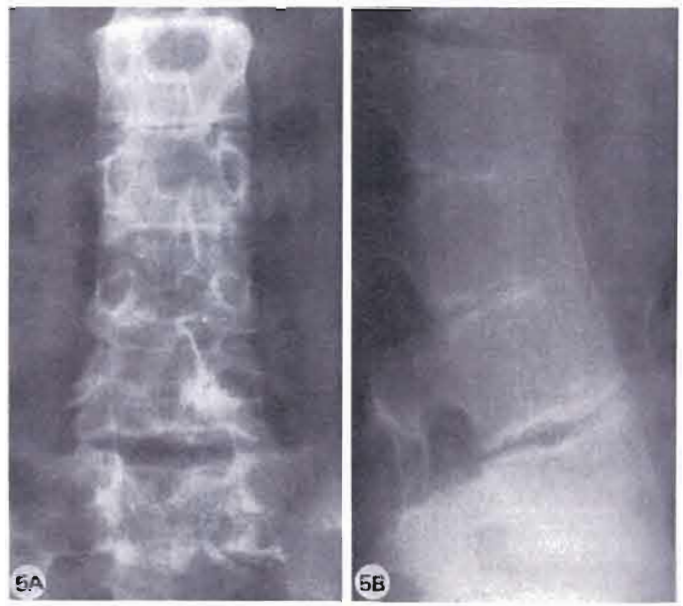

FIG. 5
A. Anteroposterior view of the lumbar spine demonstrating block vertebral segments.
B. Roentgenogram of the same patient in the lateral projection.

redistribute the formative cells into three superimposed plates, known as the primary germ layers (ectoderm, mesoderm, and entoderm). The mesoderm gives rise to most of the mesenchyme. Some of the mesenchyme may arise from ectoderm or entoderm, hence the difficulty in distinguishing the exact nature of notochordal origin (Arey, 1954). The germ layers are essentially the embryonic disc which is seated on the nutritive yolk sac. A primitive groove develops when invagination of the superficial ectodermal layer folds (Fig. 8). This will form the neural canal (Elliot *et al.*, 1970). At the base of the neural canal (on its ventral surface) a fairly elongated row of cells develops from the mesoderm below it (Källen, 1968). This tissue streak is called the notochord. If the neural canal (eventually the spinal cord) does not develop, neither does the notochotd. That the notochord is all mesenchyme is uncertain; a portion may be ectodermal or entodermal (Ehrenhaft, 1943; Källen, 1968).

The knowledge of the effects of one tissue layer on another is incomplete but the effect does exist (Cooper, 1965; Laale, 1971; Marin-Padilla, 1966). The neural plate in which the primitive groove develops is created in part, by an inductive influence from the underlying mesodermal structures. Some of the inductive influence is probably chemical (Källen, 1968). The notochord will not develop properly or at all unless the ventral spinal cord

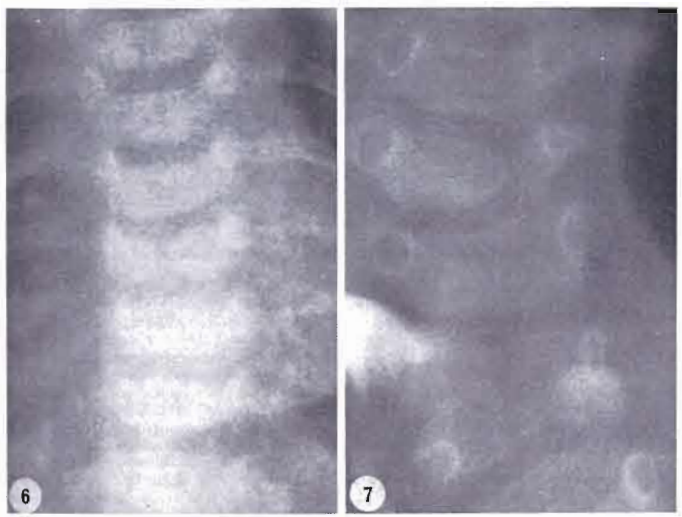

FIG. 6
A mid-dorsal butterfly vertebra. This patient had a heart anomaly (Tricuspid atresia, VSD, and a ductus arteriosus).

FIG. 7
Hemivertebrae at two levels in a patient with associated renal malformation (ectopic kidney and horseshoe). The hemivertebra may be due to hyposegmentation (the higher abnormality) or hypersegmentation (the lower abnormality).

develops from the ectoderm (Elliott *et al.*, 1970), and the surrounding mesenchyme will not develop into normal vertebrae unless the notochord develops properly (Källen, 1968; Marin-Padilla, 1966).

If the dorsal spinal cord is present, but the notochord at the same level is surgically extirpated the mesenchyme fails to develop a normal vertebral body but the arch will develop normally (Cooper, 1965). As stated above, the notochord has a direct influence on the development of the surrounding mesenchyme into vertebral bodies. With continued development of the vertebral bodies, the notochord is buried in the bone of the spinal segments and either disappears or remains vestigeal. The notochord does exert a stronger or different influence at alternate levels (intervertebral disc spaces) and disallows chondrification of mesenchyme (vertebral body formation) but helps create and becomes part of the nucleous pulposus in the intervertebral disc tissue (Elliott *et al.*, 1970). An uninhibited effect of notochord influence where vertebral chondrification and ossification generally occur can result in anterior spina bifida, butterfly vertebrae or hemivertebrae formation and/or fusion anomalies, 'block vertebrae', or congenital absence of vertebrae (Ehrenhaft, 1943; Elliott *et al.*, 1970).

Influences of the notochord are not only directed upon the lateral masses of mesenchyme (vertebral body formation). A ventral effect upon developing entodermal structures might be suspected by the relationship of the juxtaposed tissues (Elliot *et al.*, 1970). Notochordal influences may be suspected by reports of anterior spina bifida with fibrous bands connecting the exposed notochord with anomalies of the foregut (Fallon *et al.*, 1954). That is direct evidence of a physical relationship which might suggest other direct influences of one tissue layer on another. These bands of vertebral abnormalities have been reported with oesophageal cysts, duplications along the path of the primitive pharynx into the upper abdomen, and with tracheo-oesophageal diverticula (Neuhauser *et al.*, 1958).

Other Data and the Present Series. – The present series of patients was studied since the author was impressed by the numbers of references alluding to spine changes and specific somatic or visceral abnormalities. Each suggests a fundamental relationship (cause and effect) between the vertebral malformation and the somaticovisceral abnormality. There is virtually no portion of the anatomy that has not been related to the spine when congenital abnormalities of either are analysed. A consideration of all available data from this study and the literature reveals that somatic and visceral abnormalities are frequently found with congenital anomalies of the vertebrae and a definite relationship does exist. Frequently references are found to vertebral malformations and abnormalities related directly to the axial skeleton (intradural lipomas, scoliosis, diastematomyelia, intraspinal dermoid cysts and enteric cysts, lumbosacral dimples, angiomas) (Foster *et al.*, 1969; Till, 1969) and to appendicular skeletal abnormalities (usually as some syndrome) (Freedom and Park, 1973; Kirkpatrick *et al.*, 1965; Morrison *et al.*, 1968).

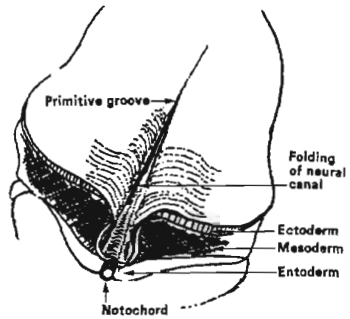

FIG. 8
A drawing depicting early embryogenesis. Discussion of the diagram appears in the text.

In the cervical spine vertebral malformations alone and in a configuration suggesting a Klippel–Feil abnormality are found with lesions of the upper oesophagus or respiratory tract and congenital heart disease, respectively. Oesophageal atresia and vertebral malformations are noted together frequently demonstrating an unequivocal but modest association. The incidence of oesophageal atresia with anomalous vertebrae varies considerably from report to report, however (Barson, 1970; Bond-Taylor *et al.*, 1973). The same holds true for tracheo-oesophageal fistulae. Although it is suggested that there is a significant correlation between tracheo-oesophageal fistulae and vertebral malformations (Stevenson, 1972) the author believes the correlative index actual and significant, but low.

Various congenital spine abnormalities have been reported in association with other gastrointestinal

malformations at levels other than the upper oesophagus (Colquhoun, 1968). These abnormalities have been reported in relation to any and every part of the alimentary tract. The most frequent are found at the cephalad (oesophageal) and caudal ends (ano-rectal) (Elliot *et al.*, 1970; Montagnini and Pampaloni, 1968; Reed and Griscom, 1973; Rosenfelder, 1968); however, the stomach and bowel are also noted in reference to dysrhaphism (Elliot *et al.*, 1970). Low cervical spine vertebral malformations and dorsal spine abnormalities are reported with duplication cysts (Elliot *et al.*, 1970; Fallon *et al.*, 1954; Moll, 1968; Neuhauser *et al.*, 1958; Rhaney and Barclay, 1959; Wilson, 1969) (respiratory, neurenteric, oesophageal, mesenteric), anomalies of the respiratory tree (tracheo-oesophageal fistulae, pulmonary aplasia, hypoplasia, dysplasia, sequestration) (Arcomano and Azzoni, 1967; Avery, 1968; Caffey, 1972; Stevenson, 1972) and congenital cardiac malformations. The author believes the coexistence of spine abnormalities with heart lesions is high, much higher than ever previously suggested. In that same range fall pulmonary aplasia and perhaps mediastinal duplication anomalies. Less directly correlated are dorsal vertebral anomalies with oesphageal and tracheal malformations.

Congenital heart abnormalities are emphasised in the present series, as stated. They are reported frequently in the literature in the Klippel–Feil syndrome (Forney *et al.*, 1966; Lynch and Grissom, 1966; Morrison *et al.*, 1968; Nora *et al.*, 1961) (a spine–heart association) and other syndromes in which the spine is also dysrhaphic (Kirkpatrick *et al.*, 1965; Ramsey and Bliznak, 1971). Interestingly, Klippel–Feil abnormalities are also reported with enteric cysts duplications, renal anomalies and other skeletal malformations (Gray *et al.*, 1964). The embryological considerations in this report yield a logical explanation for the occurrence of all of the above relationships.

On occasion thoracic vertebral abnormalities are seen in association with renal anomalies though the latter are more often related to lumbar spine malformations. The genitourinary tract malformations associated with spinal abnormalities are well known to radiologists. Attention has been called to this association by many authors (Carrig *et al.*, 1969). Suggestions of a cause and effect are notable in the discussion of some renal abnormalities and dysrhaphism. The genitourinary abnormalities include renal, ureteral, bladder and urethral problems (Smart, 1965). The most frequent renal anomaly associated with significant vertebral malformations are absence of a kidney, malpositions of the kidney, or renal malformations (horseshoe, 'cake', fusion). Although others have related ureteropelvic and ureterovesical junction obstructions to spinal abnormalities the correlation, probably valid, is low and is so in this series. The author feels because of previous, but undocumented experience, that if a large enough series were collected the true incidence would be demonstrated to be higher. Nevertheless, many more congenital visceral abnormalities (such as ureteropelvic junction and ureterovesical junction obstructions) are found without than with spine abnormalities. This is also true with all other visceral abnormalities encountered and herein discussed. The remainder of the genitourinary tract is subjected to congenital variation with associated lumbar and sacral vertebral malformations frequently evident. This includes bladder anomalies of anatomical and perhaps functional abnormality (see below) and congenital urethral malformations.

Lumbosacral abnormalities are frequently reported with imperforate anus, ano-rectal atresia, septate bladders, hydrometrocolpos, absent vagina, and small uterus, in addition to other skeletal and visceral abnormalities in the same patients. This is excellently and most emphatically documented in patients with the caudal dysplasia syndrome. That syndrome is most often seen in infants of diabetic mothers (but not always with that association) (Reed and Griscom, 1973; Rosenfelder, 1968).

The vertebral malformations referred to as 'significant' in this discussion exclude spina bifida occulta. Most likely that anatomical variant is not insignificant and there is probably some correlation between spina bifida occulta and somatic or somaticovisceral abnormalities. The problem is unresolved here as elsewhere since the exact frequency of spina bifida occulta is extremely high and variable and is directly related to the patient's age. Nevertheless, the author's feelings and actual experience, as well as those of others (Laurence *et al.*, 1968; Till, 1969) suggest that the presence of spina bifida occulta is related to somaticovisceral malformations much the same as vertebral body maldevelopment. There is a strong suggestion resulting from this study and some of the literature, that vesicoureteral reflux is seen more often with lumbar or sacral spina bifida occulta than without it and that finding cannot be accounted for on the basis of the great frequency with which spina bifida occulta is found in any one specific age group (Manley and Frech, 1970). The exact reason is undetermined. Since the presence of a vertebral malformation suggests a congenital visceral anomaly one might consider minimal

anatomical variations at the ureterovesical junction (allowing reflux); or since the arch anomaly may reflect some variation associated with a widened interpediculate distance (and an associated spinal cord abnormality) perhaps a neurologic cause for reflux exists. Certainly neurologic dysfunction of bladder and urethra have been reported as a cause for genitourinary problems (incontinence, 'bladder neck' obstruction, etc.) (Smart, 1965) and these problems may be expected or suggested if significant or even insignificant spinal changes are present. The specific quality of the spinal malformations encountered in this series and those referred to in the literature do not seem to be related to any one particular somaticovisceral abnormality. Whether the spinal lesion is a dysplasia, hypoplasia, butterfly, hemivertebra, fusion, etc., seems to be unimportant *vis-à-vis* the visceral anomalies which present. All forms are more important an indicator of the presence of congenitally abnormal viscera than spina bifida occulta. The importance of the latter is not underestimated, however, as is suggested above. There is some evidence to suggest that true spina-bifida (cystica) with symmetrical vertebral body malformation is less often associated with congenital visceral abnormalities than spina bifida with associated fusions, butterfly vertebra, etc.

CONCLUSIONS

1. There is an embryonic explanation for the association of vertebral maldevelopment and somatic and visceral abnormalities.

2. The vertebral malformations, including spina bifida occulta, are related to somaticovisceral abnormalities with varying degrees of correlation, the most frequent being congenital heart disease and renal aplasia or malposition. Less frequent, but definitely related, are oesophageal atresia, tracheo-oesophageal fistulae, mediastinal duplication cysts and pulmonary aplasia. Other associations are notable and the correlation real, but infrequent.

3. The level (location) of the vertebra or vertebrae involved may direct attention to which congenital somatic or visceral abnormality might be found.

4. The exact nature of the vertebral malformation is not an important indicator of which somaticovisceral abnormality might be found.

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