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Classification of congenital abnormalities of the sacrum

Patterns of associated dysfunctions

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Fifty-eight patients with a congenital abnormality of the sacrum were studied; a modified classification is suggested. Type 1a is characterized by an abrupt termination of the sacrum with an otherwise normal spinal column. Type 1b shows a similar sacral deficit, but with abnormalities evident higher in the spine. In Type 2 the terminal spine is malformed, and in Type 3 there is posterior sacral dysraphism. A high incidence of concomitant congenital abnormalities was found in Types 1 b and 2. Bladder dysfunction in the absence of typical clinical signs was often found in Types 1a and 1b.

Patients born with a congenital abnormality of the sacrum comprise a heterogeneous group ranging from virtually normal individuals with only a very minor sacral or coccygeal defect discovered as an incidental radiographic finding to those so severely handicapped as to require the prolonged attentions of specialists in many fields. This heterogeneity reflects not merely the degree, but also the various patterns of disability. We wish to show that within this spectrum discrete patterns may be recognized, each with its own peculiar problems.

Stanley et al. (1979) based their classification on the observations that a vertebra may fail to develop altogether, it may be malformed, or there may be posterior dysraphism resulting respectively in agenetic, dysgenetic, and dysraphic categories. These three patterns were associated with other congenital abnormalities. Urinary tract dysfunction was particularly common among the dysgenetic group. However, there is a considerable degree of overlap between the agenetic and dysgenetic groups in terms of associated congenital abnormalities and bladder dysfunction.

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A modified classification is proposed that clearly defines those patients most at risk from other structural and functional disorders.

Patients and methods

Fifty-eight patients with a congenital abnormality of the sacrum were examined at a special clinic at Alder Hey Children's Hospital in Liverpool. The patients were examined clinically, radiographically, and, where indicated, urodynamically.

By considering abnormalities in the lumbar spine, we modified our previous classification (Stanley et al. 1979) by subdivision of the agenetic group into Types 1a and 1b.

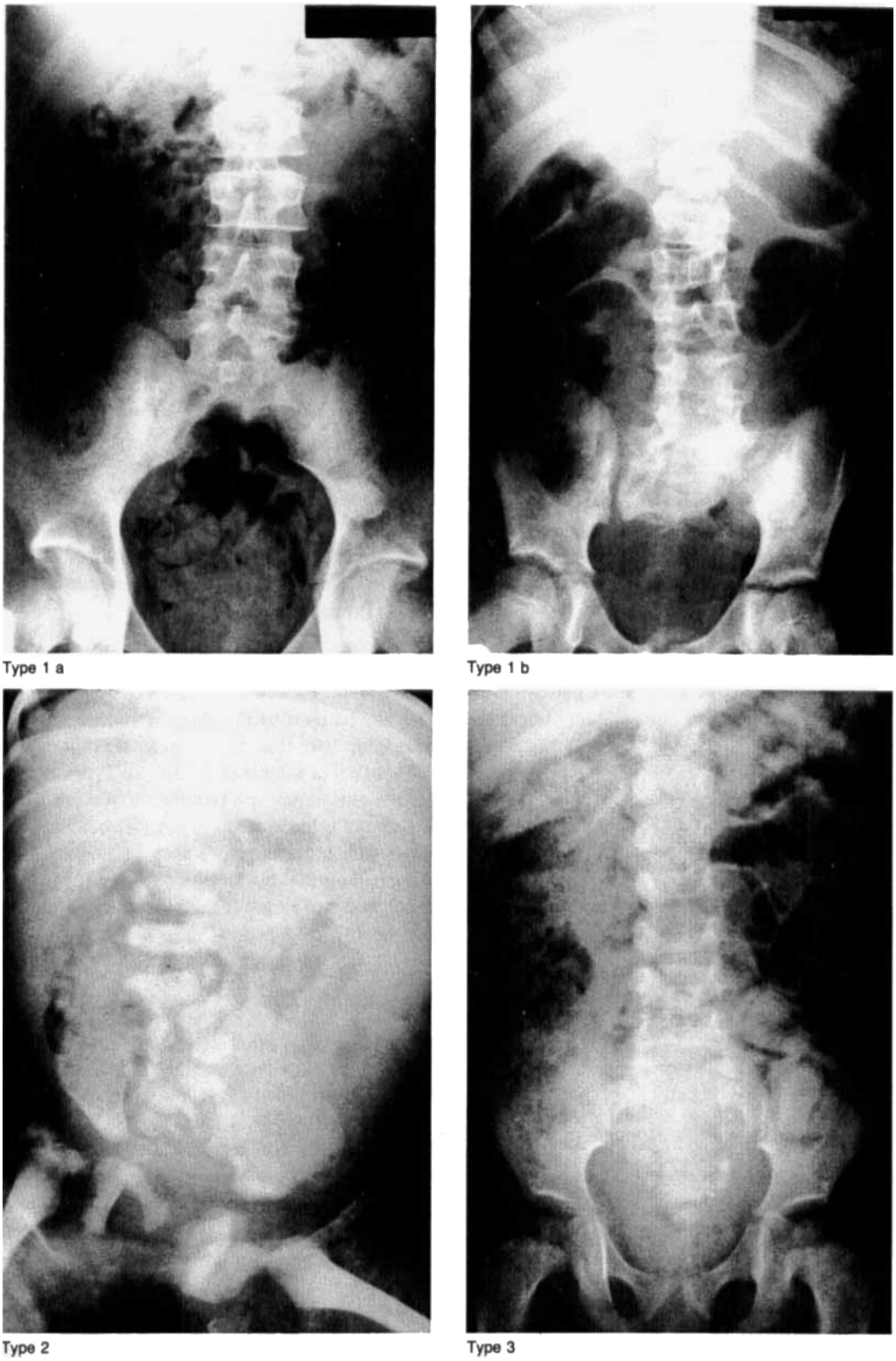
The four categories (Figure 1) were therefore as follows:

Type 1 a – 13 patients. Abrupt termination of the sacrum over no more than one segment and with the spine above the sacrum appearing normal radiographically.

Type 1 b – 12 patients. Sacral appearance as in 1a, but with congenital abnormalities demonstrated in the thoracic or lumbar spine.

Type 2 – 26 patients. The terminal vertebral elements were malformed, e.g., hemivertebrae, butterfly vertebrae, anterior spina bifida. The lumbar spine often contained abnormal elements.

Type 3 – 7 patients. Posterior spina bifida involving the sacrum.



Type 1 a

Type 1 b

Type 2

Type 3

Figure 1. Classification of congenital abnormalities of the sacrum. Also see the text.

Table 1. Malformations in patients with congenital abnormality of the sacrum

Type	n	Genito-urinary	Gastro-intestinal	Skeletal	Cardio-vascular	Other	Total
1 a	13	0	0	0	0	1	1
1 b	12	3	4	2	1	2	7
2	26	15	21	8	3	5	23
3	7	1	1	1	1	1	2

To test their validity, various clinical features were then tabulated with respect to these new categories.

Results

Congenital abnormalities (Table 1)

The abnormalities in the gastrointestinal system were mainly imperforate anus and esophageal atresia, in the genitourinary system anatomic aberrations that had usually been demonstrated on intravenous urography, and in the skeletal system true developmental failures, e.g., reduplication of long bones or lobster-claw foot.

Among the 13 Type 1a patients, a case of a minor branchial arch remnant was the only congenital abnormality. Types 1b and 2 patients both had a high incidence of concomitant congenital abnormalities.

Neurogenic lower limb anomalies

Included were such abnormalities as paralysis, claw toes, pes cavus, club foot, and leg length discrepancies that do not represent true congenital deformities. Neurogenic impairment was found in 7/13 Type 1a, in 8/12 Type 1b, in 12/26 Type 2, and in 3/7 Type 3 cases. The abnormalities correlated with the degree of neurologic loss rather than with the pattern of sacral abnormality.

Sensory sparing

This feature is often regarded as being a classical finding in "sacral agenesis." While the radiographic appearances would suggest that there should be no neurologic function beyond a given level, there was preservation of sensory function in 9/13 Type 1a, in 7/12 Type 1b, in 6/26 Type 2, but in none of the Type 3 cases. In no cases was there preservation of motor function. In 2 Type 2 cases, motor and sensory loss was greater than would be expected. Sensory sparing is thus a feature peculiar to Types 1a and 1b.

Table 2. Bladder function in patients with congenital abnormality of the sacrum

Lower limb neurology		Intact	Intact	Impaired
Bladder-function		Normal	Neuropathic	Neuropathic
Type	n			
1 a	13	2	7	4
1 b	12	4	5	3
2	26	11	4	11
3	7	1	1	5

Urologic problems

Half the patients had bladder dysfunction, many of whom had loss of perineal sensation and reduced pelvic floor muscle tone. In Types 1a and 1b patients, clinical examination was of little prognostic value in terms of bladder function. Of those with more than two sacral segments, one third had normal bladder function while most of those with two or fewer sacral segments suffered neuropathic dysfunction.

Discussion

Since Hohl (1852) first described congenital absence of the sacrum, a number of classifications have been proposed (Foix and Hillemand 1924, Durham-Smith 1959, Banta and Nicholls 1969, Renshaw 1978). Based solely upon the morphology of the lower vertebral column, these classifications fail to address the wider range of structural and functional anomalies that so often afflict these patients. Urologic dysfunction (Mariani 1979), gastrointestinal abnormalities (Berden et al. 1966, Williams and Nixon 1957), and congenital abnormalities elsewhere in the body often cause considerable morbidity.

Patients with a congenital abnormality of the sacrum suffer principally from their neurologic deficit and associated congenital abnormalities. Stanley et al. (1979) stated that concomitant congenital abnormalities and bladder problems occur principally in the dysgenetic group, as do abnormalities higher in the spine. Yet, in our series we noted that all of these features were also found in certain members of the agenetic group. On the basis of vertebral abnormalities in the thoracic and lumbar spine, we subdivided the agenetic Type 1 into two, Types 1a and 1b. This resulted in the identification of those patients whose features had blurred the distinction between the agenetic and dysgenetic groups. Another important finding was that sensory sparing was found to be a feature only of the agenetic category, the neurologic deficit in Types 2 and 3 correlating well with the skeletal deficit.

The sacrum is the earliest portion of the body to differentiate, and the motor and sensory components of the cord are separate during their initial stages of development. With this in mind, it might be concluded that an early teratogenic insult could lead to failure of development of the lowermost motor elements and corresponding vertebral segments while the sensory and autonomic elements, should they have already received their organizing stimulus, would continue to develop normally. If the insult is brief, further development higher in the spine and organogenesis elsewhere should resume normally. The embryo will then have a loss of terminal vertebral segments with a corresponding motor loss, but with normal sensory function and few congenital abnormalities elsewhere, constituting the Type 1a patient. Should the insult take a little longer to abate, while the caudalmost pattern would be as before, development higher in the spine would then also be disrupted along with organogenesis elsewhere in the body that is now well under way – the Type 2 patient. The Type 3 patient is likely to occur as a result of a similar process to that which produces spina bifida higher in the spine, probably a blockage in cerebrospinal fluid flow results in ballooning of the cord and of the overlying posterior vertebral elements with neurologic loss corresponding to the bony deficit. This event occurs towards the end of the first trimester when organogenesis is nearly complete, so the incidence of concomitant congenital abnormalities is relatively low.

This hypothetic mechanism for the differing patterns of deformity is based upon the chronologic timing of the insult and makes no mention of the nature of the teratogen. Many teratogenic agents have been shown to have the capacity to produce sacral abnormalities in experimental animals, but that which receives most frequent attention in the literature is maternal diabetes, with a reported incidence as high as 16 percent (Blumel et al. 1969, Passarge and Lenz 1966). In our series, 2 patients, both Type 1a, were born to diabetic mothers. Quite why diabetes leads to fetal malformation has been a matter of some debate, but it seems that the teratogenic factor is hyperglycemia (Baker et al. 1981). The insult leading to the Type 1a malformation, as postulated above, would of necessity be brief in duration, well furnished by a hyperglycemic episode in a mother whose diabetic control becomes somewhat unbalanced by early pregnancy.

The importance of classifying these patients lies in the early identification and treatment of associated problems. While certain anomalies will be self-evident, urinary function in particular may lead to irreversible renal damage in the presence of only subtle symptoms particularly in the very young child. By identifying the exact pattern of sacral abnormality, the efforts of the clinician may be focussed on those areas most likely to yield further pathology.

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