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Special report

Instability of the upper cervical spine

SKELETAL DYSPLASIA GROUP*

There are probably about 100 different types of skeletal dysplasia (osteochondrodystrophy), most of them genetic in origin and associated with growth retardation and short stature. Many, in addition, have premature degeneration of joints and childhood osteoarthritis.[1] Localised structural bony defects are less common, but odontoid absence has been known to occur occasionally. Interest and alarm has also been expressed at the high incidence of atlantoaxial instability in Down's syndrome, variously reported at between 10 and 20%, and its association with serious neurological complications after minimal trauma.[2–4]

The Skeletal Dysplasia Group at a 1985 meeting (independently of these latter reports) discussed the problems connected with odontoid hypoplasia or absence in generalised disorders of the skeleton and planned a largely retrospective review of cervical spine anomalies in all patients on their register, where feasible obtaining up to date radiographs with flexion/extension views. This paper contains a short description of the anatomical problems and a report of findings from seven skeletal dysplasia clinics in Britain.

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Anatomy and measurement of cervical instability

The normal anatomy at the base of the skull, with the atlas and axis, is shown in figs 1–4. The main

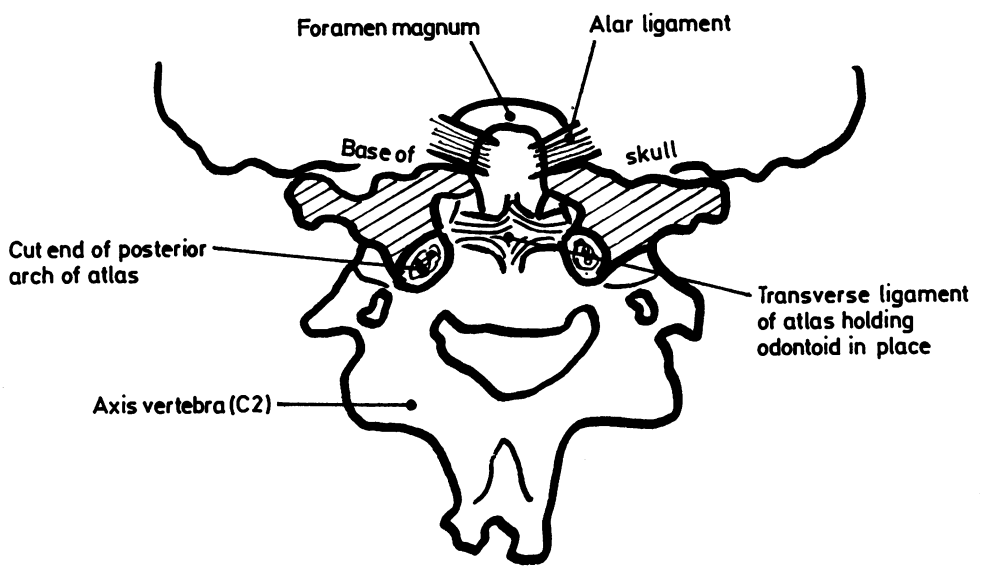

Fig 1 *Posterior aspect of the upper cervical spine with the posterior arch of the atlas cut away to show the transverse and alar ligaments. In Morquio's disease not only does the odontoid fail to develop but the ligaments are deficient, softened, and inefficient.*

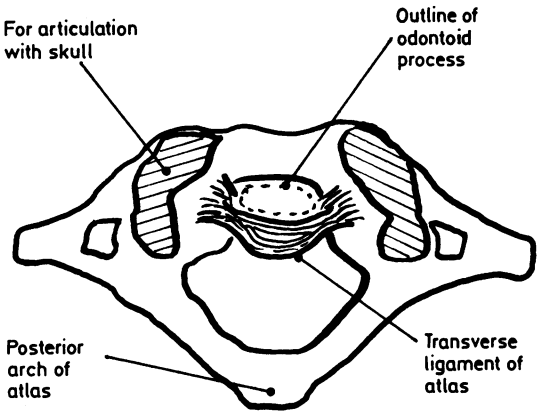

Fig 2 *View from above showing the position of the odontoid process in relation to the anterior arch and transverse ligament of the atlas.*

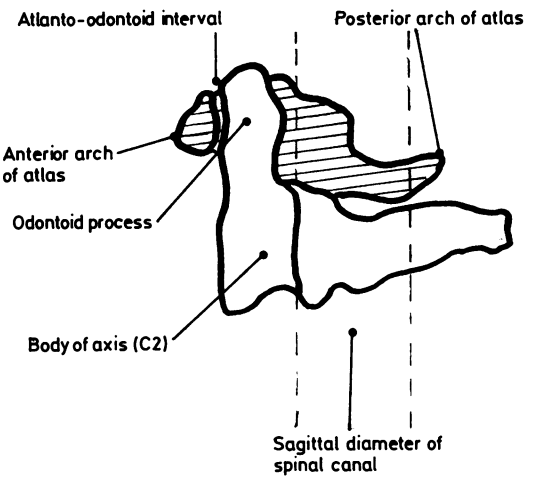

Fig 3 *Diagram of the lateral aspect of the atlas and axis in the neutral position showing the two most important measurements relating to stability: the atlanto-odontoid interval and the anteroposterior canal diameter (sagittal diameter)—see text.*

features ensuring stability of the region, while allowing movements of the head and neck to take place, are (1) the bony structure of the axis with its odontoid process; (2) the transverse ligament of the atlas: a thick, strong band which holds the odontoid process in contact with the anterior arch of the atlas; and (3) the alar ligaments of the odontoid process: two strong, rounded cords arising one on each side of the upper part of the odontoid process, passing upwards obliquely and laterally to be inserted into the occipital bone.

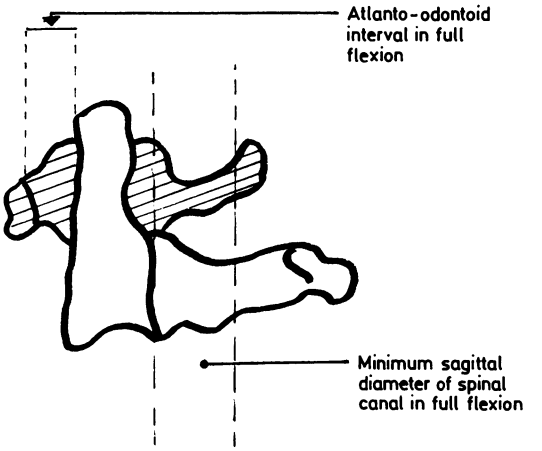

Fig 4 *In full flexion the atlanto-odontoid interval increases and the canal diameter decreases. The narrowing of the spinal canal is obvious compared with fig 3. Increased movement on flexion and extension occurs when there is instability in this region, whether associated with a fractured, hypoplastic, or absent odontoid, or deficient ligaments, or more than one of these factors (see figs 6–7).*

Although vertebral anomalies may be noted on plain radiographs, this does not necessarily mean there will be spinal instability endangering cord function. Lateral radiographs in flexion and extension are necessary in order to carry out measurements.[5 6] The atlanto-odontoid interval is the distance between the posterior edge of the anterior arch of the atlas and the front of the odontoid process (or, in its absence, a line projected upwards from the front of the body of C2). In normal children up to the age of 7 years the measurement should not exceed 4·5 mm, but in adults, with a fully ossified skeleton and in whom ligaments are less elastic, it is only 2·5 mm.[5]

The second important measurement is the minimum anteroposterior canal diameter (sagittal diameter). This is the distance between the posterior border of the body of the axis and the posterior arch of the atlas, measured in full (voluntary) flexion. It is difficult to gain cooperation from small children, and precise figures are not available for them, but Greenberg states that if this distance is 14 mm or less then cord compression always occurs; if between 15 and 17 mm it may occur, but compression virtually never occurs if the diameter exceeds 18 mm.[5]

Subjects and results

There were 182 patients with osteochondrodystrophies on the register for whom adequate cervical

Table *Upper cervical anomalies in the skeletal dysplasias*

	No of patients	*Age range (years)*	*No with odontoid hypoplasia*	*No with absence*	*No with other defects: base of skull to C3*	*No with known neurological complications*	*Total No (%) with bony defect*
Morquio's disease	15	1–15	7	8	0	5	All 15 (100)
Other mucopolysaccharide disease:							
Hurler's	6	1–13	4	0	4	0	15 of 20 (75)
Hunter's	5	3–14	3	0	0	0	
Scheie's	3	6–14	1	0	1	0	
Sanfilipo's	3	1–11	3	0	0	0	
Maroteaux Lamy's	3	3–6	3	0	0	0	
Spondyloepiphyseal dysplasia:							
congenita	24	Neonate–54	15	1	4	1	20 of 24 (83)
tarda	18	3–62	3	0	1	1	4 of 18
Pseudoachondroplasia	23	Neonate–53	8	0	5	0	13 of 23 (57)
Achondroplasia and hypochondroplasia	23	2 months–65	1	0	1	0	2 of 23
Multiple epiphyseal dysplasia	18	4–adult	6	0	0	0	6 of 18
Chondrodysplasia punctata	14	Neonate–11	2	0	9	0	11 of 14
Metatropic dysplasia and Kniest disease	7	Neonate–15	1	1	2	0	4 of 7
Diastrophic dysplasia	6	Neonate–48	1	0	3	1	4 of 6
Spondylometaphyseal dysplasia	3	7–16	1	0	0	0	1 of 3
Total	171		59	10	30	8	80 (47)

spine radiographs were available; findings for 171 of them are shown in the table. Eleven patients with sclerosing bone dysplasias or tumour like disorders such as osteopetrosis and diaphyseal aclasis showed no abnormality and are not discussed further. Osteogenesis imperfecta was excluded as problems in this area of the spine are compounded by osteoporosis and possible basilar invagination. No patient with Down's syndrome had presented at the skeletal dysplasia clinics. Patients with malformation syndromes were also excluded.

It is clear that not only odontoid dysplasia but other anomalies from the base of the skull to the third cervical vertebra are not unusual among this group of diseases (48% in total). Twenty six flexion extension radiographs were available for study, but the only positive signs of instability were found in 15 patients with Morquio's disease, all of whom were either frankly unstable or borderline, as judged by the measurements described above (figs 5–7). Five of these 15 had known neurological complications and some had already undergone surgery for stabilisation.

Cord compression in the cervical region had occurred in only three other patients: one with diastrophic dysplasia and two with spondyloepiphyseal dysplasia, one with the cogenita form and the other tarda, the latter patient being 62 years of age before problems occurred. We are suspicious that the sudden and unexpected death of one other adult patient with spondyloepiphyseal congenita, after a road traffic accident, was also due to this cause, but no postmortem examination was performed.

The incidence of upper cervical anomalies without known neurological problems is surprisingly high—particularly in spondyloepiphyseal dysplasia congenita (83%) (fig 8a and b), the mucopolysaccharide disorders other than Morquio's disease (75%), and pseudoachondroplasia (57%). The proportion is probably high also in diastrophic and metatropic dysplasias and Kniest disease, although few cases with adequate radiology were available in these extremely rare disorders.

MORQUIO'S DISEASE AND EARLY PRESENTING SIGNS OF NEUROLOGICAL DAMAGE

Deterioration begins usually during the middle years of childhood (4–9 years of age), but early neurological signs are not immediately and easily differentiated from existing mechanical problems in the lower limbs that are associated with severe joint laxity and often gross genu valgum. At this age, these patients should be reviewed every three months and recent physical deterioration noted: parents will be aware of the child's loss of endurance, tiredness, 'going off his feet', and perhaps fainting attacks. It is not unusual for symptoms to arise from

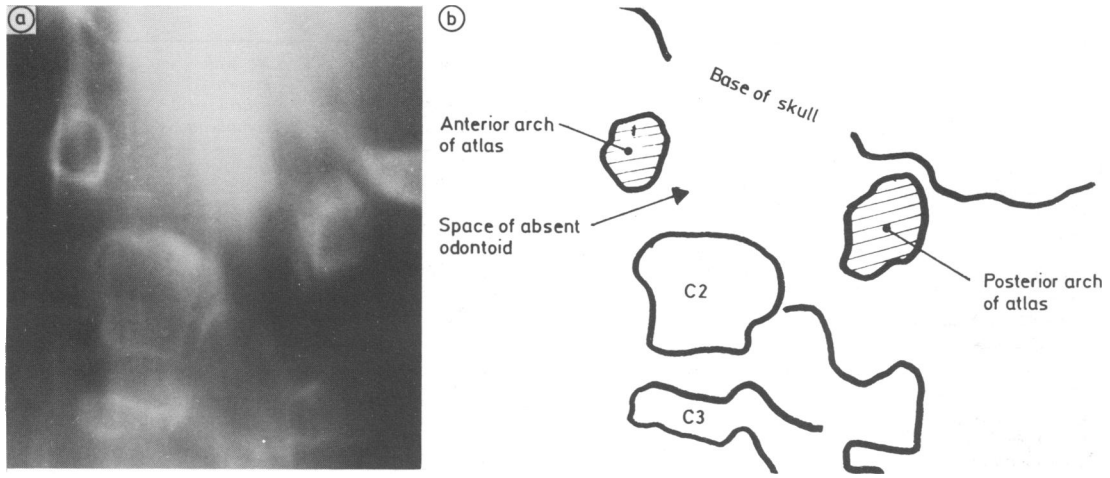

Fig 5a and b *Tomogram and tracing of the lateral aspect of the cervical spine (neutral position) in a child with Morquio's disease, indicating the space which should be occupied by the odontoid process.*

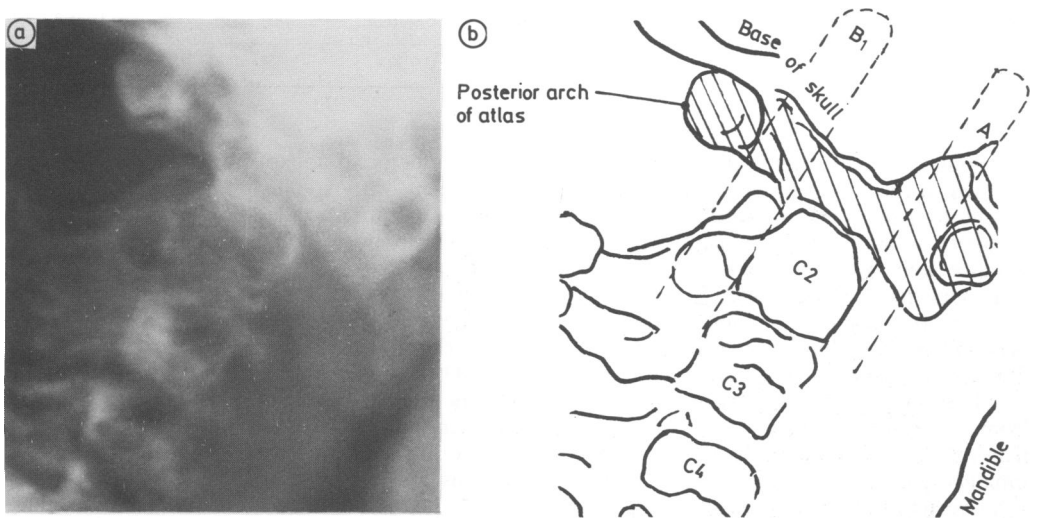

Fig 6a and b *Radiograph and tracing of flexion view in Morquio's disease. The atlanto-odontoid interval (A) is increased (6 mm on the original radiograph)—that is, the body of the axis is too far back, impinging on and causing appreciable narrowing of the anteroposterior canal diameter (B_1).*

lower down the cervical cord, with 'shocks' of 'pins and needles' going down the upper limbs. The position of the head may be significant, being held tilted backwards, in extension, where 'it feels more secure'—as indeed it is (figs 7a and b). The patient may perhaps refer to a fear of the head 'falling off'. Damage (which is irreversible) to the respiratory centre of the medulla will manifest itself as sleep apnoea or sensitivity to low oxygen concentrations—for example, at high altitudes or during anaesthesia. Long tract signs come later and urinary signs later still.

The authors are not all agreed on this point, but as serious and irreversible neurological damage occurs with great frequency in Morquio's disease, there is a case to be made for taking early flexion/extension views of the cervical spine and carrying out prophylactic stabilisation, perhaps around 6–8 years of age. Surgery at an earlier age would be even more hazardous than it already is, with the child's excess

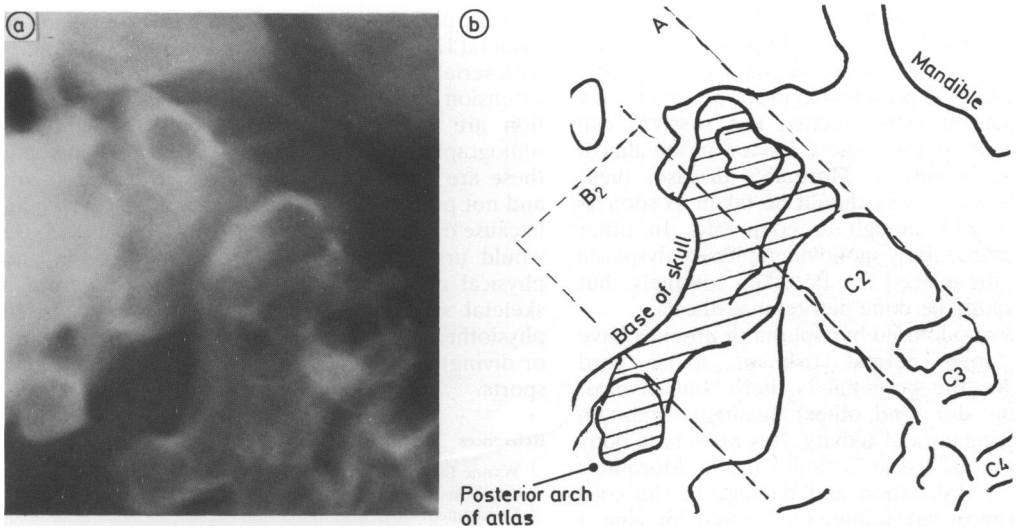

Fig 7 a and b *Radiograph and tracing of extension view in Morquio's disease. The body of the axis has now moved too far forward and lies in front of the anterior arch of the atlas (A); consequently the anteroposterior canal diameter is much increased (B_2 and refer to figs 6a and b). It is obvious why a child with cervical instability must hold his head permanently in extension.*

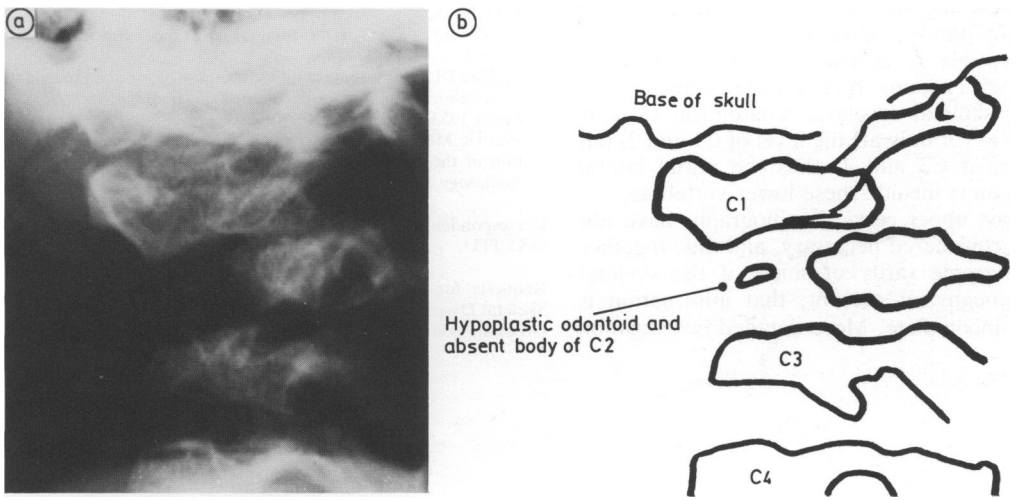

Fig 8a and b *Lateral radiograph and tracing of the upper cervical spine in a patient with spondyloepiphyseal dysplasia congenita. The anterior part of the body of the axis is hypoplastic, the odontoid process absent, and there is appreciable subluxation.*

of cartilage and the consequent difficulty in obtaining bony fusion. The area of anatomy described in fig 1 occupies only about one square inch in these tiny children: it is difficult surgery in a dangerous area. Skilled anaesthesia is crucial, and the procedure should be carried out in centres experienced in the technique.

Discussion

There is clearly a high incidence (48%) of upper cervical anomalies in a variety of skeletal dysplasias. Probably all patients with generalised developmental disorders of the skeleton should have a lateral radiograph of the cervical spine to establish whether

or not there are abnormalities here. In a child this region can usually be included on the lateral radiograph of the skull. Observation of a hypoplastic or absent odontoid is useless in itself, however; only flexion/extension views (active not passive) can show instability in this region. In view of the almost invariable problems in Morquio's disease, these flexion/extension views should be taken as soon as the child is old enough to cooperate. In other diagnoses, particularly spondyloepiphyseal dysplasia congenita, these need not be taken routinely, but should certainly be done before anaesthesia.

All cases of odontoid hypoplasia or absence have a higher than normal risk of spinal cord compression—the potential is there, but in most cases strong alar (and other) ligaments maintain stability during normal activity. It is when both bony and ligamentous tissue is faulty (as in Morquio's disease) that dislocation and damage to the cord becomes almost inevitable. One could imagine a similar situation arising in other 'joint laxity' conditions, such as an Ehlers-Danlos syndrome, if by chance the odontoid process were absent or fractured. It is also likely that patients with other skeletal dysplasias are at risk when reduced mobility of joints, including those of the neck, are associated with upper cervical anomalies, as they will then be more susceptible to damage in this region from relatively minor trauma. It must also be remembered that in the skeletal dysplasias anatomical variants may not be at (or only at) the level of C1 and 2, but lower down at C3 and 4, thus the initial lateral radiograph must include these lower vertebrae.

In the past upper cervical radiographs have not often been considered necessary, and this, together with the extreme rarity of some of the skeletal dysplasias means, at present, that information is scanty and incomplete. More detailed investigation of these patients is required and is continuing in the Skeletal Dysplasia Group by the authors and others, with serial studies and more frequent use of flexion/extension radiographs. Other methods of investigation are available: air myelography,[7] computed tomography, or magnetic resonance imaging, but these are usually for evaluating selected problems and not practical for routine screening. Meanwhile, because of this potential risk to the cervical cord, we would urge caution both during anaesthesia and physical activity for any person suffering from a skeletal dysplasia. Walking, cycling, supervised physiotherapy, and swimming (but not underwater or diving) are recommended, but not body contact sports.

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