Congenital spinal deformity: assessment, natural history and treatment

Peter R Loughenbury

Nigel W Gummerson

Athanasios I Tsirikos

Abstract

Congenital spinal deformity is caused by vertebral anomalies that produce an imbalance in spinal growth. Anomalies can be classified as failures of vertebral formation, failures of segmentation or a mixture of both. The natural history and risk of deformity progression depends on the anatomical location and type of anomaly. Patients should be assessed with a thorough clinical evaluation including investigations for renal, cardiac and intraspinal anomalies. Timely diagnosis and estimation of risk of progression is required to allow early surgical intervention before a severe curve develops. This may take the form of *in situ* fusion or hemi-epiphysiodesis where there is minimal deformity. In moderate deformities a posterior instrumented correction and fusion may be needed, while in severe deformities osteotomies or vertebral column resection may be required. Surgery in patients with congenital spinal deformity carries a risk of neurological injury so an early simple surgical treatment should be favoured to minimize the risk of complications.

Keywords congenital kyphosis; congenital scoliosis; surgical treatment; VEPTR; vertebral anomaly

Introduction

Congenital spinal deformity comprises a spectrum of sagittal and coronal plane deformities that are caused by vertebral anomalies – including scoliosis, kyphosis and kyphoscoliosis. Asymmetrical failure of development in utero leads to imbalanced growth and spinal curvature that is present at birth and can continue until skeletal maturity.[1,2] The anatomical pattern of this failure of vertebral development can lead to slow or rapid progression of the deformity and this produces a range of clinical pictures. These range from a benign curve with low potential to deteriorate to a more malignant curve that can progress rapidly and cause cardiorespiratory, functional, neurological and cosmetic complications.

Prevalence of congenital spinal deformity is approximately 1 in 1000 live births.[3] Congenital scoliosis is the most common deformity (80%) followed by congenital kyphoscoliosis (14%) and isolated congenital kyphosis (6%).[4] The cause of the deformity is often not clear and is likely to be multifactorial. Maternal exposures to diabetes, carbon monoxide, anti-epileptic drugs, and alcohol have been implicated as possible causative factors.[5–7] Genetic inheritance has been shown for some vertebral anomalies and these are often identified as part of an underlying syndrome such as Klippel–Feil, Alagille, Goldenhar, Jarcho–Levin and VACTERL (vertebral, anal atresia, cardiac, tracheo-oesophageal fistula, renal and limb anomalies).[8]

Embryological development of the spine[9–11]

Embryological development of the spine and ribs occurs during the fourth to sixth weeks of gestation. During this period the mesenchymal anlage is established and gives rise to the vertebral and rib growth centres. Failures of formation or segmentation can occur at this stage and if unilateral can lead to an imbalance of spinal growth. Cartilaginous and ossification stages follow as the vertebral levels become more distinct and vertebral anomalies can arise during both these stages. Failure of vascularization can occur in the developing cartilage and lead to a failure of formation, whilst bony metaplasia can occur in the annulus or ring apophysis during the ossification phase and produce an unsegmented bar (failure of segmentation).

The ribs form from lateral outgrowths of the mesenchymal anlage (costal processes). In the thoracic spine these develop into cartilaginous precursors that ossify during the fetal period. Rib anomalies occur due to failures of segmentation of the developing somites in the thoracic spine, as due to the vertebral abnormality the ribs cannot articulate with a definitive thoracic vertebra.

Classification

Congenital deformities of the spine can be classified according to their anatomical location and the pathological anatomy of the anomaly. They can either be due to a failure of vertebral formation or segmentation, and can occasionally be due to a mixture of both of these processes (Figure 1).

Failures of formation occur when there is absence of part of a vertebra. This can be complete or incomplete. In a *complete failure* of formation one half of the vertebra fails to form and this leads to a hemivertebra. This can be fully segmented (disc spaces and end plates above and below the hemivertebra so that it is separated from the adjacent levels), semi-segmented (fused to one caudal or cephalad vertebra with a fully formed disc space separating it from the other), unsegmented (fused to the bodies above and below and therefore without any growth potential) or incarcerated (a small ovoid vertebral segment between the caudal and cephalad levels with limited potential to grow). In both segmented and semi-segmented hemivertebrae there are normal growth

Peter R Loughenbury *MSc PG Cert Med Ed FRCS (Tr & Orth) Consultant Orthopaedic Spine Surgeon and Honorary Senior Lecturer, Leeds Centre for Neurosciences/University of Leeds, Leeds General Infirmary, UK. Conflicts of interest: none declared.*

Nigel W Gummerson *MA FRCS (Tr & Orth) Consultant Orthopaedic Spine Surgeon, Leeds Centre for Neurosciences/University of Leeds, Leeds General Infirmary, UK. Conflicts of interest: consultancy agreements with K2M and Stryker.*

Athanasios I Tsirikos *MD FRCS PhD Consultant Orthopaedic and Spine Surgeon and Honorary Clinical Senior Lecturer, University of Edinburgh/Royal Hospital for Sick Children, Scottish National Spinal Deformity Centre, Edinburgh, UK. Conflicts of interest: educational/teaching commitments with DePuy/Synthes Spine.*

Please cite this article in press as: Loughenbury PR, et al., Congenital spinal deformity: assessment, natural history and treatment, Orthopaedics and Trauma (2017), https://doi.org/10.1016/j.mporth.2017.09.007

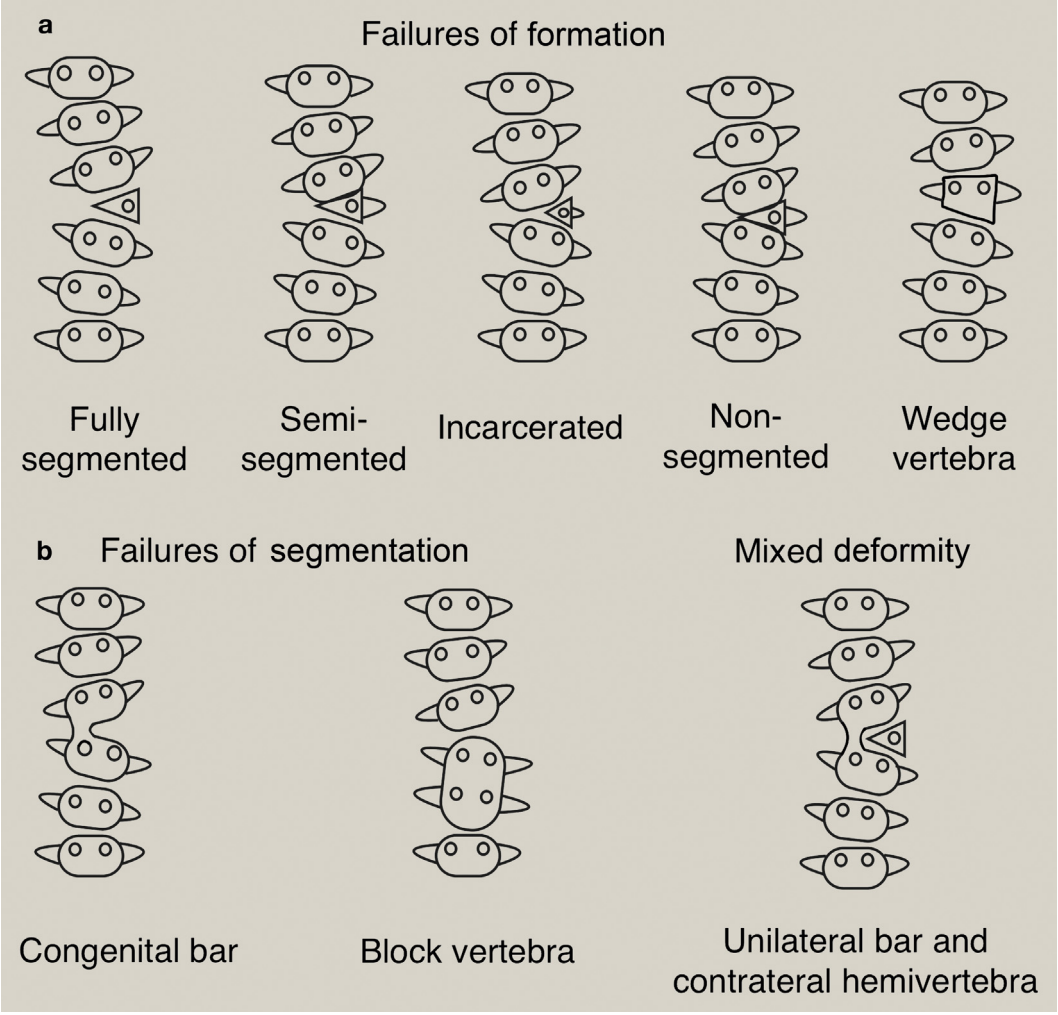

Figure 1 Classification of congenital scoliosis: (**a**) failures of formation; (**b**) failures of segmentation and mixed anomalies.

plates on one side of the spine and this can lead to asymmetrical growth and the development of a spinal deformity. An *incomplete failure* of formation includes a wedge vertebra and this can also produce asymmetrical growth. A more pronounced sagittal plane deformity can be seen where the failure of formation produces a posterolateral quadrant vertebra (formed due to antero-lateral vertebral aplasia) and a subsequent kyphoscoliosis. A pure kyphosis might occur if there was a posterior hemivertebra caused by symmetrical anterior aplasia of the vertebral body. Where there are multiple hemivertebrae these may occur at ipsilateral adjacent or non-adjacent levels, which lead to more pronounced asymmetrical growth and the development of scoliosis. Conversely, a hemivertebra may be counterbalanced by a hemivertebra on the contralateral opposite side in the same anatomical region. When there is at least one normal vertebra in between this is termed '*hemimetameric shift*' – a phenomenon that is most commonly seen in the thoracic region.[12]

Failures of segmentation occur when there are abnormal connections between vertebrae. These connections form a bar between vertebral segments which produces a tethering effect that can span across two or more levels. Scoliosis can occur as the result of imbalanced growth due to the contralateral growth centres and can be more severe in the presence of one or more contralateral hemivertebrae. If the bar is bilateral then a block vertebra is formed. This produces no asymmetrical growth and has a low risk of causing a significant global deformity.

Mixed anomalies contain both failures of vertebral formation and segmentation within the same patient. These patients may have a greater risk of deformity progression due to the cumulative effect of the malformations. The spinal deformity is often associated with chest wall abnormalities that can affect the growth of the spine and the thoracic cavity. It can be very difficult to identify the nature of mixed anomalies at birth because the spine is only 30% ossified at this stage. Deformities can also produce an '*aligned*' or '*displaced*' spine. A displaced deformity occurs where there is a subluxation or dislocation of the column adjacent to a vertebral anomaly and this has a greater risk of curve progression and a more significant risk of neurological complications.

These classification systems concentrate on the anatomy of the anterior column due to the potential for asymmetric growth when there is an anterior deformity. However, surgical

Please cite this article in press as: Loughenbury PR, et al., Congenital spinal deformity: assessment, natural history and treatment, Orthopaedics and Trauma (2017), https://doi.org/10.1016/j.mporth.2017.09.007