

Goldenhar Syndrome (Oculo-Auriculo-Vertebral Spectrum) and the Spine

A patient guide

In brief: Goldenhar syndrome, more precisely known as the oculo-auriculo-vertebral spectrum (OAVS) and overlapping closely with hemifacial microsomia and craniofacial microsomia, is a congenital condition arising from disrupted development of the first and second pharyngeal (branchial) arches early in pregnancy. Its name draws together its three classic domains: the eye (oculo-), the ear (auriculo-) and the spine (vertebral). It is the third element, the vertebral component, that brings the condition into the orbit of spinal surgery. Although the facial and ear differences are usually the most visible features, a substantial proportion of those affected also carry anomalies of the spine, particularly of the cervical and upper thoracic vertebrae. These range from minor, symptomless variations of bone shape through to failures of vertebral formation and segmentation that can produce congenital scoliosis, fusion of the neck vertebrae, instability at the junction between the skull and the spine, and, occasionally, narrowing around the spinal cord. This page concentrates on that spinal dimension: what the vertebral anomalies are, how often they occur, how they are recognised, and what the published literature reports about their natural history and management.

What Goldenhar syndrome is, and why the spine is part of it

Goldenhar syndrome sits at the more complete end of a continuous spectrum of conditions that share a single underlying theme: incomplete or asymmetrical development of structures that arise from the first and second pharyngeal arches during the early weeks of embryonic life. These arches give rise to much of the face, jaw and ear, and disturbances in the migration and survival of the cranial neural crest cells that populate them are thought to lie at the heart of the condition. Because the same developmental window governs the formation of the early spinal segments, the somites, it is unsurprising that the bones of the spine are drawn into the same process.

The terminology can be confusing, and it helps to set it out plainly. Hemifacial microsomia describes the underdevelopment of one side of the face. Craniofacial microsomia is a broader umbrella term for the same family of features. Oculo-auriculo-vertebral spectrum, or OAVS, names the wider constellation that adds eye and vertebral involvement, and Goldenhar syndrome is the historical eponym, often reserved for cases that combine the facial microsomia with epibulbar dermoids of the eye, ear tags or appendages, and vertebral anomalies. In practice these labels overlap heavily and are frequently used interchangeably, and the spinal aspects described here apply across the spectrum.

The condition is not rare in absolute terms among congenital craniofacial differences. Reported incidence figures vary considerably according to how the diagnosis is defined, but commonly cited estimates fall between roughly 1 in 3,500 and 1 in 5,600 births, with some series citing figures as wide as 1 in 26,000. A modest male predominance is usually described. Most cases are sporadic, occurring without a clear family history, although a minority show inheritance patterns and several genes, including SF3B2, FOXP1 and PAX1, have been implicated in some families. From the spinal point of view, the genetics matter chiefly because they underline that the vertebral anomalies are present from birth, are congenital in origin, and are not the result of later wear or injury.

The vertebral component: how common, and where in the spine

The "vertebral" in oculo-auriculo-vertebral is not a token addition. Vertebral anomalies are one of the defining domains of the spectrum, although the reported frequency varies widely between studies, partly because different groups have imaged the spine to different extents and used different definitions.

Across the literature on craniofacial microsomia, the reported prevalence of vertebral anomalies ranges enormously, from around 8 per cent to nearly 80 per cent. A large retrospective analysis of nearly a thousand patients found vertebral anomalies in around 28 per cent, while studies that focus specifically on the more complete Goldenhar phenotype, and that image the spine systematically, tend to report higher figures, with some describing spinal abnormalities in the majority of those examined. When the cervical region is studied in detail, fusion of vertebrae has been reported in a high proportion of patients. The headline message is consistent even if the precise number is not: a clinically meaningful share of people with the condition carry an anomaly somewhere in the spine.

The distribution within the spine is characteristic. The cervical and upper thoracic, or cervicothoracic, region is by far the most frequently involved, which mirrors the developmental link between the spine and the affected pharyngeal arches. The base of the skull and the first few cervical vertebrae attract particular attention, because malsegmentation here, often with diffuse fusion between the skull base and the upper neck, has been described as the most common spinal pattern in tomographic studies of Goldenhar syndrome. The lower thoracic and lumbar spine are involved less often, although they are not exempt, and individual case reports describe unusual lumbar anomalies. A notable feature is that the side of the facial underdevelopment tends to correspond to the side of the cervical and craniocervical abnormalities, so the asymmetry of the face is, in a sense, echoed in the spine.

Congenital scoliosis in the oculo-auriculo-vertebral spectrum

Of all the spinal manifestations, scoliosis is the one most relevant to long-term spinal health, and it is among the most frequently reported. The scoliosis seen in this setting is, importantly, a congenital scoliosis: a curve that arises because the vertebrae themselves formed or divided abnormally, rather than the more common adolescent idiopathic scoliosis that appears in an otherwise normally built spine.

Congenital scoliosis is conventionally grouped by its underlying bony fault. Failures of formation occur when part of a vertebra never develops, the classic example being a hemivertebra, a wedge-shaped half-vertebra that tilts the spine to one side. Failures of segmentation occur when adjacent vertebrae fail to separate, producing a block of fused bone or an unsegmented bar down one side that tethers growth on that side while the other side continues to lengthen. Mixed patterns, combining both faults, are also common. Each of these underlies a curve that behaves differently from an idiopathic one, because the deforming force is built into the architecture of the bone and is driven by the asymmetry of growth.

In Goldenhar syndrome the scoliosis is most often cervicothoracic in location, frequently accompanied by torticollis, a tilting and turning of the head, which itself reflects the asymmetry of the upper spine and neck. Thoracolumbar curves and kyphoscoliosis, where a forward bending is combined with the sideways curve, are described less often. Because congenital curves are present from birth and are tied to growth, their behaviour over time depends heavily on the specific bony anomaly: some are stable and progress little, whereas others, particularly unsegmented bars opposite a hemivertebra, are recognised in the wider congenital scoliosis literature as among the most likely to worsen during periods of rapid skeletal growth. The presence of hemifacial microsomia among children with congenital scoliosis is itself reported, in one large series, in around 2 per cent of cases, illustrating the two-way link between the conditions.

Cervical vertebral anomalies: fusion, Klippel-Feil and the neck

The neck is the heartland of spinal involvement in this spectrum. Failures of segmentation in the cervical vertebrae produce fusions, where two or more vertebrae are joined into a single block of bone. When multiple cervical vertebrae are congenitally fused, the pattern is termed Klippel-Feil, classically associated with a short neck, a low posterior hairline and restricted neck movement, and Klippel-Feil-type fusions are a recognised association of the oculo-auriculo-vertebral spectrum.

The functional consequence of these fusions is a reduction in the range of movement of the neck, sometimes considerable, because motion that would normally be shared across several mobile joints is concentrated into the

remaining unfused segments. This concentration of movement is part of why fused necks attract long-term interest: the segments adjacent to a fused block carry more load and motion than they otherwise would. In the published series, however, it is worth noting that only a minority of patients with vertebral anomalies report symptoms from them, and the commonest of those reported symptoms are torticollis, neck or back discomfort, and limited neck movement rather than anything more serious.

A further consideration in the upper neck is the relationship between the cervical anomalies and the airway. Fusion and malsegmentation of the upper cervical vertebrae, together with occipitalisation of the atlas, reduce neck mobility and can complicate the positioning of the head, a point that is given particular weight in the anaesthetic literature and is discussed further below.

The craniocervical junction: occipitalisation, the odontoid and instability

The junction where the skull meets the spine, the craniocervical or craniovertebral junction, deserves separate attention because anomalies here have the greatest potential to matter. Several distinct findings are described in the oculo-auriculo-vertebral spectrum.

Occipitalisation of the atlas, also called atlanto-occipital assimilation, is a congenital fusion of the first cervical vertebra to the base of the skull, so that the atlas becomes, in effect, part of the occiput. This is a recognised association of the spectrum and of Klippel-Feil syndrome. Hypoplasia, or underdevelopment, of the odontoid peg, the upright projection of the second cervical vertebra around which the head rotates, is also reported. Basilar invagination, where the top of the spine sits higher than normal relative to the skull base, completes the picture of craniocervical anomalies that have been documented in this condition.

The clinical significance of these findings lies in their potential to affect stability and, more rarely, the spinal cord. Instability at the C1-C2 level, the atlantoaxial joint, has been described as a frequent finding in the spectrum. The literature notes that such instability may be silent in childhood yet may progress as skeletal maturity approaches, which is one reason the craniocervical region receives careful and repeated attention. In the great majority this remains a radiological observation rather than a source of neurological trouble, but documented cases exist in which craniocervical instability, sometimes alongside other anomalies such as a tethered spinal cord, has required surgical attention. These remain uncommon, and the literature describing them is largely confined to individual case reports and small series.

Other spinal and adjacent skeletal findings

Beyond the curves, fusions and craniocervical anomalies, several other findings round out the spinal picture. Block vertebrae, where two vertebral bodies are congenitally united, are among the commonest individual anomalies reported. Spina bifida occulta, a minor and usually symptomless failure of the bony arch at the back of a vertebra to close fully, is frequently noted on imaging and is generally of no clinical consequence in itself. Rib anomalies, including missing, fused or extra ribs, often accompany the thoracic vertebral anomalies, reflecting the shared developmental origin of vertebrae and ribs.

Anomalies of the shoulder girdle, in particular an elevated and underdeveloped scapula known as Sprengel deformity, are associated with the same family of conditions and with Klippel-Feil, and contribute to the overall asymmetry of the upper trunk. Although these are not strictly spinal, they sit alongside the vertebral findings and form part of the broader skeletal expression of the spectrum. Their relevance to the spine is mainly that they may coexist with, and add to the appearance of, an underlying congenital curve or cervical anomaly.

How the spinal anomalies are recognised and assessed

Because the vertebral anomalies are congenital and bony, their assessment rests on imaging, and the literature emphasises systematic spinal imaging in those diagnosed with the spectrum, particularly given that a curve or fusion may be present without producing any symptoms.

Plain radiographs of the whole spine provide the overview, allowing curves to be measured, fused or block vertebrae to be identified, and the overall balance and alignment to be judged. Because the anomalies cluster in the neck and cervicothoracic junction, dedicated views of this region are often part of the assessment. Computed tomography, the CT scan, with its detailed depiction of bone and its ability to be reconstructed in three dimensions, is described in the literature as particularly informative for clarifying the precise nature of vertebral malsegmentation and the craniocervical anatomy, and tomographic studies have been central to characterising the distinctive spinal patterns of Goldenhar syndrome. Magnetic resonance imaging, the MRI scan, complements this by showing the spinal cord itself, which is relevant where craniocervical anomalies, basilar invagination or a suspected tethered cord raise the question of whether the neural structures are affected.

A recurring theme across the literature is that early and systematic identification of congenital vertebral anomalies allows spinal deformity to be recognised at an early stage, and several authors describe routine referral of those with the spectrum for spinal assessment during growth. Surveillance over time, rather than a single assessment, is emphasised, because congenital curves and craniocervical findings can change as the skeleton grows and matures. These are descriptions of the approaches reported in the published literature and not recommendations directed at any individual.

What the evidence shows about management

The evidence base for managing the spinal aspects of the oculo-auriculo-vertebral spectrum is built largely on case reports, small retrospective series and the broader congenital scoliosis literature, rather than on large trials. The descriptions that follow report what that literature observes, and decisions in practice are always individualised within a specialist team.

For the many anomalies that are minor, stable and symptomless, such as an isolated block vertebra, spina bifida occulta or a non-progressive curve, the literature describes observation and periodic monitoring rather than active treatment. Many cervical fusions and minor vertebral variations fall into this category and are reported to remain asymptomatic over long periods.

For congenital scoliosis, the published approach mirrors that of congenital scoliosis in general. Where a curve is mild and not progressing, monitoring through growth is described. Bracing has a limited and debated role in congenital curves because the deformity is structural, although the literature includes the occasional report of a curve responding to bracing. Where a curve is clearly progressive, particularly one driven by a hemivertebra or an unsegmented bar, the surgical literature describes a range of techniques. In younger children with substantial remaining growth, growth-friendly approaches, including rib-based and spine-based expandable systems, have been reported as effective in this population. In suitable cases, removal of a hemivertebra combined with a short fusion is described as a means of correcting the local deformity, and longer fusions are used for established curves. These are technical descriptions drawn from published series, not guidance for any particular person.

For craniocervical anomalies and instability, the literature describes monitoring of stable findings and reserves stabilising surgery for the uncommon situations where instability is significant or progressive, or where the spinal cord is threatened. The general principle running through the surgical literature is that intervention is matched to the severity and progression of the problem, and weighed against the recognised risks of operating in this anatomically complex and often asymmetrical region.

Surgical and anaesthetic considerations specific to the spine

Two features of the spectrum interact in a way that is given particular emphasis in the surgical and anaesthetic literature, because they bear directly on the safety of any procedure, including spinal surgery.

The first is the airway. The facial and jaw underdevelopment characteristic of the condition, especially mandibular hypoplasia, is well recognised to make securing the airway for general anaesthesia difficult, and the difficulty is reported to correlate with the severity of the jaw underdevelopment. The cervical anomalies compound this,

because fusion, malsegmentation and occipitalisation limit the neck movement that would ordinarily help with positioning for intubation. The anaesthetic literature therefore stresses careful preoperative airway assessment and attention to a controlled, neutral neck position throughout, particularly where the cervical spine may be unstable.

The second is the heightened risk profile of major spinal surgery itself in this group. A recent analysis reported that patients with Goldenhar syndrome undergoing spinal deformity surgery had increased odds of respiratory failure, readmission and reoperation compared with others, and the authors suggested that targeted measures to reduce these risks may be appropriately considered. The respiratory dimension is amplified where thoracic vertebral and rib anomalies have affected the development and capacity of the chest. Taken together, these considerations explain why the literature consistently frames spinal surgery in this population as a matter for careful multidisciplinary planning, with the airway, the cervical spine and the chest all weighed alongside the deformity itself.

Outlook

The outlook for the spine in Goldenhar syndrome is, for most people, considerably more reassuring than the long list of possible anomalies might suggest, and it tracks closely with the distinction between minor, stable findings and progressive deformity. Many of the vertebral anomalies, including isolated fusions, block vertebrae and spina bifida occulta, cause no symptoms and require no treatment, and even among those who do have vertebral anomalies only a minority report any symptoms from them, most commonly torticollis, neck or back discomfort, or limited neck movement.

The findings that carry the greatest weight are progressive congenital scoliosis, where the asymmetry of vertebral growth drives a worsening curve, and significant craniocervical instability, where the junction between skull and spine is affected. Both are uncommon, both are the focus of monitoring through growth, and both have established, if individualised, pathways of management when they do arise.

Across the whole field, the consistent themes are that the spinal anomalies are congenital and present from birth, that they cluster in the neck and upper thoracic spine and tend to mirror the side of the facial difference, that the majority are minor and stable, and that the small number which progress are best identified early through systematic assessment. Any individual outlook is a matter for evaluation by the treating specialist team rather than something that can be read from a diagnosis or a category alone.