HEMIVERTEBRAE, ARE THEY ALWAYS SYMPTOMATIC?

Lorna Jackson¹  Vivek Chandra²  Colin Michie³

ABSTRACT

Most hemivertebrae eventually lead to congenital scoliosis. We describe an infant with an asymptomatic hemivertebra that occurred as part of a VACTERL syndrome. The effects of hemivertebrae, incidence, detection, risks, treatment and prognosis of congenital scoliosis, are outlined.

Competing Interests: None

Key Words: Hemivertebra, Scoliosis, congenital scoliosis, Vacterl

CASE HISTORY

LJ is a Caucasian girl with known VACTERL syndrome. At the age of 8 months she was admitted with recurrent episodes of respiratory distress, fever and coryzal symptoms. She was hypertensive (115/80). Nasopharyngeal aspirate showed parainfluenza virus type 3. Blood culture was negative. Her treatment was supportive, with intravenous fluids and face-mask oxygen. Since birth, LJ has had five hospitalizations. She has had infections including RSV bronchiolitis necessitating CPAP ventilation, and empirical antibiotic coverage. She has required intubation on two occasions because of hypoxemia and hypercapnia. She is currently on medication to control her hypertension, prophylactic antibiotics for urinary tract infection, and nebulised saline to assist with physiotherapy.

LJ was born at 38 weeks gestational age by elective Cesarean Section, the second infant in the family. She required resuscitation and intubation at birth. She received surgery on day 2 of life to correct a tracheo-oesophageal fistula.

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She was treated for severe reflux with a Nissen Fundoplication at which time a percutaneous endoscopic gastrostomy (PEG) was placed to assist with feeding. Investigation after surgery showed a number of features (table 1) consistent with a VACTERL association. LJ has some dysmorphic features with frontal bossing and a short neck. She has an XY karyotype with normal chromosomal banding. On this admission, she made an uneventful recovery and was discharged home after 5 days. LJ’s hemivertebra is in the thoracic region, and therefore probably has a reasonable prognosis with respect to her spine. At present no obvious spinal deformity is noted on physical examination. She has an appointment to see the orthopedic specialist.

Table 1: Features of LJ consistent with VACTERL syndrome.

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<thead>
<tr>
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<td>Renal ultrasound</td>
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<td>MRI Brain</td>
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<td>EEG</td>
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<td>Echocardiogram</td>
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<td>Chest X-ray</td>
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DISCUSSION

EFFECTS OF HEMIVERTEBRA

LJ’s hemivertebra is located in the upper thoracic spine. Congenital vertebral malformations generally occur in early embryonic life (before 7 weeks), and are thought to represent errors in formation or segmentation of the spinal segments that originate from primitive mesenchymal condensations of embryonic cells. (1) This congenital malformation may result in either congenital scoliosis or congenital kyphosis depending on whether it was a lateral hemivertebra, or a dorsal hemivertebra respectively (Fig 1). Scoliosis is a lateral curvature of the spine with vertebral rotation. LJ’s hemivertebra was a lateral hemivertebra which indicates that she will develop a congenital scoliosis.
Congenital scoliosis is caused by one of two types of structural bony abnormality. Type I is a failure of formation, such as that seen with hemivertebrae (Fig 2 A-E). Type 1 may give rise to wedge vertebra or butterfly vertebra. Type II is a failure of segmentation, such as that seen with block vertebrae and that seen with unsegmented bars, where growth is tethered on one side of the spine (Fig 2 F-G). Unilateral unsegmented bars with contralateral hemivertebrae have the most powerful tendency to rapid progression and should be surgically fused as soon as the bony abnormality is evident.(2). The degree of scoliosis produced depends on four factors: the type of the hemivertebra; its site, the number of hemivertebrae and their relationship to each other; and finally, the age of the patient (3).

INCIDENCE AND DETECTION OF HEMIVERTEBRA

Hemivertebra is seldom seen at birth and is clinically difficult to detect unless a radiograph.(4) LJ had difficulty breathing with grunting and flaring of the nostrils the chest x-ray showed hemivertebra in the upper thoracic region. Ultrasound has been used in the prenatal diagnosis of hemivertebra. (5) In a study done over a three year period, six cases of hemivertebra were diagnosed with ultrasound between fourteen and twenty-three weeks gestational age.(6) A comparative study on diagnosis of fetal dysplasia of
spine and spinal cord between ultrasonography and Magnetic Resonance Imaging (MRI), showed MRI to be a valuable complement to sonography in difficult cases, and diagnostic accuracy can be significantly improved. (7).

About 1:10,000 in the USA is affected with congenital scoliosis, and over the years this number have remained constant (10). In a study done at a tertiary medical center in Israel, of 78,500 live births, 26 (0.33/1,000), were found to have hemivertebra. The ratio for male/female was 1:1. Twenty-three out of the 26 infants (88.5%) with hemivertebra had additional congenital anomalies (cranial, cardiac, renal, intestinal, and skeletal).(8)

Figure 2: Types of Congenital Scoliosis (2)
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RECURRENT RISK

Isolated hemivertebrae in siblings is very uncommon, although an association of hemivertebrae with neural defects in siblings has been reported. Wynne-Davies performed a family survey on 337 patients with congenital scoliosis and found that 5 to 10% of sibling of patients with multiple vertebral anomalies with or without spina bifida had either vertebral anomalies or spina bifida. However, she found only one of 245 siblings of 101 infants with a solitary vertebral defect (including isolated hemivertebra) had a spinal defect and concluded that isolated defects were sporadic (non-familial) in nature and carried no risk to subsequent siblings. Connor et al. found an incidence of 4% of neural tube defects among siblings with congenital scoliosis. In contrast to the previous report, they found an increased incidence of neural tube defects in siblings of probands with single hemivertebra as well as those with multiple vertebral defects Therefore, it would be reasonable to offer genetic counseling and prenatal diagnosis for neural tube defects to those patients with a previous child with vertebral anomalies. (9)

ASSOCIATIONS OF HEMIVERTEBRA

Most fetuses with prenatally diagnosed hemivertebrae have additional anomalies, often syndromic, which affect the prognosis. Hemivertebra may be part of syndromes including Jarcho-Levin, Klippel-Fiel, and VACTERL (9), of which VACTERL is the most frequent. VACTERL Association is a nonrandom association of vertebral defects (eg hemivertebra), anal atresia (imperforate anus), cardiac defects (eg VSD), tracheoesophageal fistula, esophageal atresia, renal defects, and limb defects (radial).(10). LJ is a known VACTERL association and, as well as having a thoracic hemivertebra, includes an association of the following abnormalities:

<table>
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<tr>
<th>Cardiac</th>
<th>Patent Ductus Arteriosus, and Patent Foramen Ovale</th>
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<tr>
<td>Tracheal and Oesophageal</td>
<td>Tracheo-oesophageal fistula</td>
</tr>
<tr>
<td>Renal</td>
<td>Agenesis of right kidney and ectopic single left kidney</td>
</tr>
<tr>
<td>Limbs</td>
<td>Anomaly of right radius and thumb in the form of right radius and thumb shortening</td>
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Hemivertebra may be seen in children with chromosomal abnormalities, such as Trisomy 18 or Edward’s Syndrome or chromosome deletions. (11).
PROGNOSIS

Overall prognosis is not related to the spine but depends on associated anomalies. However isolated hemivertebra, carries good prognosis. Only about 1-2% of congenital scoliosis will get better by themselves. Another 2.5% stay right where they are and never require treatment, though these curves will need monitoring with x-rays to make sure they've stabilized. Approximately 75% deteriorate and require treatment (4). With respect to progression, hemivertebrae have a variable prognosis. It depends on whether a contralateral hemivertebra is present that results in overall balance of the spine, whether multiple hemivertebrae are on one side of the spine, and how much growth potential is predicted for each endplate of the hemivertebra. Hemivertebrae at the cervicothoracic junction, and the lumbosacral junction have a relatively poor prognosis because the spine above or below the abnormality cannot compensate. Hemivertebrae should be observed so as to delineate their growth potential and progression. (2)

TREATMENT FOR CONGENITAL HEMIVERTEBRA

The treatment of congenital scoliosis differs from that for idiopathic scoliosis. In congenital scoliosis, bracing is seldom effective; the bones themselves are crooked or deformed and a brace on the outside simply can't affect a discrepancy on one side or the other of the vertebral column. The only treatment that works is surgery (4) If surgical intervention in patients with congenital scoliosis is indicated, ie the 75% who do need corrective treatment, several options are available. Fusion in situ is the simplest procedure. For very young (less than 10 years) patients, a posterior fusion alone results in tethering of the posterior elements while the anterior elements continue to grow. This situation may lead to the crankshaft phenomenon, whereby the anterior growth in the spine results in a twisting deformity around the fused posterior elements. For this reason, combined anterior and posterior fusion is usually recommended for very young patients, halting growth circumferentially about the spine.(2)

In some cases of hemivertebra, epiphysiodesis may be performed; arresting growth on the curve convexity but permitting continued growth on the curve concavity, with resultant gradual curve correction. This procedure has good results in selected patients but can be unpredictable with respect to the amount of actual correction that can be achieved.(8)

In cases in which a hemivertebra is accompanied by significant coronal decompensation and compensatory growth would not be adequate to result in spinal balance, consideration can be given to hemivertebra excision via a combined anterior and posterior approach. Although this procedure is technically more demanding and has greater potential risks, it allows for better overall curve correction and improvement of coronal balance (2).
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OUTCOME

Vertebral fusion will affect future growth, but surgery at the right age will result in a longer spine (4). Since a portion of the spine is being fused, and there is no insertion of rods or wires that can help to stabilize the spine, the patient will need to wear post surgical brace or cast for protection.

Addressing congenital scoliosis at an early age will prevent deformity and pain in adulthood. In adults, cardiopulmonary considerations are frequently the source of anxiety among physicians and patients. Spirometric pulmonary function tests are usually unaffected in the idiopathic scoliosis patients until the curve exceeds 60 to 65°, and the mortality is unaffected until the curve exceeds 90 to 100°. The subjective dyspnea experienced by patients with otherwise normal arterial blood gases, can be explained by the diminishing compliance of the thoracic cage as the scoliosis increases, therefore, the work required in respiration increases. (12)

CONCLUSION

A patient with VACTERL syndrome is described; she has a hemivertebra in the upper thorax. This was not detected antenatally. As is common in the majority of such infants, involvement of other organ systems in the VACTERL syndrome is causing significant medical problems. LJ suffers from recurrent infection and possibly aspiration; at present no problems relating to her hemivertebra are detectable.

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