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Case Report

Bifurcation of the fourth rib as a possible indicator of Gorlin's syndrome in an 85-year-old female cadaver

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Abstract

We report the case of an 85-year-old female cadaver with unilateral bifurcation of the fourth rib. Causes of rib bifurcation are not well understood but have been reported in syndromic diseases such as Gorlin's Syndrome, a genetic abnormality manifesting in multiple basal cell carcinomas. The presence of Gorlin's Syndrome could not be verified in the current case though other criteria associated with the condition were present including multiple brain tumors and renal cysts. This case report supports the previous recommendations that the detection of bifid ribs warrants further investigation of other internal malformations as an important part of early diagnosis of Gorlin's syndrome, possibly reducing the severity of the long-term sequelae of the condition.

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Key words [bifid rib] [renal cysts] [brain tumor] [Gorlin's syndrome]

Introduction

Anatomical variations of human ribs have been well documented [1–3]. Common variations include asymmetric ribs, cervical or lumbar ribs, fusion of rib 1 and 2 and the absence of rib 1 [2, 3]. Unusual variants include fibrous tissue replacing costal cartilage, ribs bifurcated at the sternal insertion, two adjacent ribs articulating at their costal cartilages by a diarthrosis, synarthrosis or sydesmosis, the concurrent presence of cervical and lumbar ribs and the presence of coccygeal ribs [1].

The prevalence of rib variations has been reported to be between .04 and 14% with bifurcated ribs reported as the least common and cervical ribs as the most common [1, 2]. In another study of 47 patients with identified rib variations, the most common variant was fusion (72%), followed by bifurcation (28%) and hypoplastic ribs (26%). Vertebral variations associated with the identified rib were found in 72% of the patients [4].

The purpose of this case report is to describe a rare anatomical bifurcation in the right 4th rib in a female cadaver found during routine dissection and discuss possible underlying causes for this variant based other characteristics found in the donor. We conclude that the rib bifurcation seen in the current case combined with of other potential indicators including renal cysts and a history of brain tumors may indicate the presence of Gorlin's Syndrome in this donor.

Case Report

The single unilateral bifurcation of the right 4th rib was found in an 85-year-old female cadaver (Figures 1, 2). The donor had a history of breast cancer and cause of death was stated as a result of a brain tumor. Dissection of the skull revealed evidence of multiple craniotomies. Also noted in the donor's medical history were an appendectomy, hysterectomy, cholecystectomy, a fractured hip and brain surgery. Additionally, the presence of multiple cysts on both kidneys was noted during dissection (Figure 3).

During routine dissection of the thoracic cage a single unilateral bifurcation of the right 4th rib was found. A single sternal insertion was noted, analogous with the sternal insertion of the left 4th rib. The bifurcation occurred approximately 5 cm from the sternal insertion within the costochondral cartilage. Costochondral joints were present on both the superior and inferior portions of the bifid rib. The superior costochondral joint occurred approximately 1 cm from the medial point of bifurcation. The inferior

Figure 1. Bifurcation of the fourth rib found in a cadaveric specimen. **a)** Anterior view; **b)** Lateral view – specimen supine; **c)** Anterior view of posterior thoracic wall demonstrating costal articulations to vertebral column. *(**2, 3, 4, 5**: rib number)*

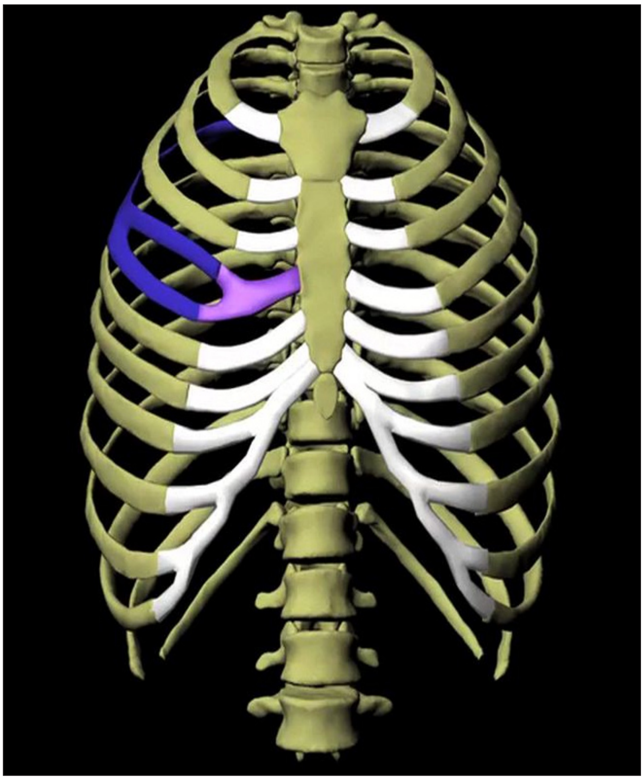

Figure 2. Three-dimensional computer model of the bifurcation of the fourth rib. QuickTime movie is available at ***http://www.ijav.org/2013/ijav_2012-056_fig-2_movie.mov***

Figure 3. Renal cysts evident on kidney of cadaveric specimen with bifurcation of the fourth rib.

costochondral joint occurred approximately 1.3 cm from the medial point of bifurcation. Both divisions of the bifurcation became bony after the costochondral joints; total length of the bifurcation was approximately 7.6 cm. The width of the 4th rib lateral to the bifurcation was considerably wider than any other rib within the thoracic cage. Articulation of the 4th rib at the costovertebral joints was noted to be normal with the head of the 4th rib articulating normally with the 3rd and 4th thoracic vertebra. No abnormalities were seen in the articulating vertebrae.

No intercostal muscles or vessels were found within the bifurcation, however a single layer of fascia was present superficial to the rib. A single vein was noted along the posterior inferior portion of the fascia. Normal neurovascular structures were found in both the 3rd and 4th intercostal spaces above and below the bifurcation. During bilateral comparison, the left 3rd and 4th intercostal spaces were wider than the right (Table 1).

Discussion

There is little information throughout medical literature regarding the clinical significance of bifid ribs. However, the presence of a bifid rib has been suggested as warranting further evaluation for nevoid basal cell carcinoma syndrome [4, 5]. Nevoid basal cell carcinoma syndrome, also known as Gorlin's Syndrome (OMIM 109400), has been well documented and is associated with the occurrence of bifid ribs [4–7]. This autosomal dominant condition displays high penetration and variable expression [7]. Estimated incidence of Gorlin's syndrome varies between 1/57,000 [8] and 1/256,000 [7]. Major manifestations include the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaw, hyperkeratosis of the palms and soles and intracranial ectopic calcifications [9]. Minor manifestations may include congenital skeletal conditions (such as bifid, splayed, fused, or missing ribs), medulloblastoma, lymphomesenteric cysts, cardiac or ovarian fibromas, renal malformation and cysts or facial dysmorphism (macrocephaly, cleft lip/palate, and severe eye abnormalities) [7, 9]. As many as 60–70% of patients diagnosed with Gorlin's syndrome demonstrate skeletal variations, commonly including the ribs, [5, 10] with as many as 26% demonstrating bifid ribs [11]. Previous research has shown an increased potential for brain tumors in individuals with a family history of Gorlin's syndrome due to the high penetrance of the condition [5]. Additionally, 5–10% of patients with Gorlin's syndrome develop the brain malignancy medulloblastoma, which may be a potential cause of early death [7].

Gorlin's syndrome is associated with somatic mutation affecting Ptch 1&2 (patched, ptc, gene) that encodes a transmembrane receptor as well as a splice mutation of SUFU both affecting sonic hedgehog pathways and somite formation. Huang et al. [12], utilizing quail chick chimeras, reported that ribs have a sclerotomal origin with proximal portions of the ribs arising from the medial somatic mesoderm while the lateral compartment gives rise to the distal ribs [13]. It would appear that the mutation in this patient was highly specific and it possibly manifested as a somatic mutation subsequent to initial rib condensation consistent with the mechanism proposed for this mutation [14]. Unfortunately, genomic DNA was not available to confirm this hypothesis.

The presence of Gorlin's syndrome in the present case study may be indicated by the presence of several minor criteria, specifically the presence of a bifid right 4th rib, a history of brain tumors and the presence of apparent cysts on both kidneys. Diagnosis of Gorlin's syndrome was not indicated in the donor's medical history and could not be verified by genetic testing due to the preserved condition of the cadaveric tissue. However, the presence of the bifid rib and other minor criteria in this case serve as an example of the potential value of bifid rib detection as an important part of early diagnosis of Gorlin's syndrome [4, 5]. The discovery of such minor manifestations should warrant further clinical examination and thus may reduce the severity of the long-term sequelae of Gorlin's syndrome [4, 5].

Table 1. Anatomic dimensions of bifurcated rib and surrounding structures.

Feature	Right Side	Left Side
3rd rib width	1.1 cm	2.0 cm
4th rib width	2.8 cm (lateral to bifurcation) 2.2 cm (medial to bifurcation)	1.4 cm
5th rib width	1.6 cm	1.6 cm
Sternum to bifurcation	5.0 cm	–
Bifurcation to costochondral joint	Superior portion: 1.0 cm Inferior portion: 1.3 cm	–
Sternum to costochondral joint	–	5.2 cm
Length of bifurcation	7.6 cm	–
Total width of bifurcation	3.7 cm (outer width) 1.4 cm (inner width)	–

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