Bifurcated rib as major diagnostic criteria of Gorlin syndrome in paediatric patient: a rare case report

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ABSTRACT

**Background:** Bifurcated rib or Bifid rib is considered to be an anatomical overgrowth anomaly of the chest wall, forming a forked sternal end. Diagnosis of this condition is generally an incidental finding on imaging or post-mortem examination. Most of the cases were asymptomatic and established as part of Gorlin syndrome. Here we describe a rare case of chest wall deformity in a paediatric patient, and our approach for this case with limited cost and resources.

**Case Presentation:** A 5-year-old boy, presented at orthopedic outpatient clinic with complaint of a painless lump in his upper left chest. No significant physical findings of patient showed, a condition caused by the mutation of PTCH1. By the time this report was being written, there was no clinical importance of isolated bifid rib yet.¹²

This study aimed to inform and educate doctors about the value of early OS diagnosis and continued observation of adolescent patients until the designated age.

**Keywords:** rib disorder, bifurcated rib, Gorlin syndrome.

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**Conclusion:** There is currently no data on the best approach to managing bifid ribs. In addition, genetic testing for susceptible genes should be strongly considered. In this situation with limited cost and resources, we suggest close monitoring with regular follow-ups for the patient as the best approach, including detection of any other diagnostic criteria of Gorlin syndrome, to ensure better prognosis and quality of life for this patient.

**CASE PRESENTATION**

A 5-year-old boy, presented by the parents at the orthopedic outpatient clinic of a general hospital with a chief complaint of a painless solid lump in his upper left chest since 1 month ago. There was no history of fever, malaise, weight loss, cough, chest pain, breathlessness, and hemoptysis. No past history of tuberculosis and other infections, malignancy or any congenital syndromes. His family history for breast cancer or any other malignancy and congenital syndromes was insignificant. The patient had no history of any trauma in the past.

The physical findings of patient showed no visible deformity of face or limbs. His head circumference was 51 cm, which was normal for his age. On examination, we found a hard palpable painless lump at the level of left 3rd and 4th intercostal space, with size of approximately 2 × 3 cm, while merging into the adjacent rib. There were no clinically significant findings in the other side of the chest or axilla. Computed Tomography (CT) of the chest revealed a bifid left 4th rib at the sternal end (Figure 1 and 2).

Since the patient was asymptomatic with no other remarkable findings, no treatment was needed. The patient is currently doing well. We suggested the family to do a genetic testing on the patient, particularly PTCH1 gene, to rule out Gorlin syndrome, a condition which may be the underlying cause of his complaint, and to rule out any predisposition to various cancerous and noncancerous tumours. However, due to unavailability of this test in Bali, Indonesia, and high-cost issues, the family refused to pursue further investigation of this case. Hence,

**BACKGROUND**

A bifurcated Rib or bifid rib is a congenital deformity of the chest wall, which are divided into structural and numerical categories. The numerical abnormalities are based on an extra or missing rib, while the structural ones are based on bifurcated, fused, hypoplastic, forked or bridging ribs. Bifurcated Rib, or commonly known as bifid rib is considered to be an anatomic overgrowth anomaly of the chest wall growth, usually present since birth. Bifid rib is often unilateral and forms a forked sternal end.¹²

This abnormality is known to be found in 1.2% of population, making up 20% of congenital chest wall deformities. Bifid rib usually involves the third and fourth ribs. Unlike any other rib malformations, bifid rib is known to occur in absence of vertebral deformities. This diagnosis is generally found as an incidental finding on X-Rays, chest CT-Scan, or post-mortem examinations. Most of the cases are asymptomatic and were known as one of the findings in Gorlin syndrome, one of the findings in Gorlin syndrome, are asymptomatic and were known as such.
a regular follow-up every four to six months has been recommended to ensure no new complications or symptoms, and appearance of other signs occur in the future. This would comprise of regular check-ups for vertebral fusion, postnatal development of macrocephaly and other related musculoskeletal anomalies, with complete skin and dental examinations, eye check-up, abdominal ultrasonography, and any related conditions at least until the age of 30 years old.

DISCUSSION

Bifid rib is one of the congenital musculoskeletal anomalies found in the ribs, where the third and fourth ribs are the most place to be affected by this condition. This rare anatomical variant is an incidental founding which is discovered in chest X-Ray, CT-Scan or in post-mortem autopsy reports. This developmental disorder caused the sternal end of a rib is split into two, unilaterally without any vertebral defect. It has been hypothesized that the abnormality of mesoderm development during embryogenesis may cause this condition due to faulty chondrification. This anomaly is found in 1.2% of population, which makes up the 20% of congenital chest wall deformities. Bifid rib is also known to be found more in men on the right side of the ribs. A retrospective study for congenital rib abnormalities in 650 patients showed that 6.76% (44 cases) of the patients were affected by bifid rib with a mean age of 21.1 ± 4.9 years and a male predilection (82%).

Bifid rib itself is mostly asymptomatic and the diagnosis alone doesn't have any significant effect in health, but in previous studies it is associated with Gorlin-Goltz syndrome. By the time this report is being made there are no significant information that discusses this specific diagnosis, but the clinical importance of previous study suggests the need of close observation and thorough examination to rule out Gorlin syndrome.

Gorlin-Goltz syndrome or as known as Gorlin syndrome, basal cell nevus syndrome (BCNS) is a rare autosomal dominant familial disorder. It is characterized by numerous basal cell carcinomas (BCCs) and ophthalmologic, neurologic, and skeletal abnormalities that usually arise from childhood. The prevalence of Gorlin syndrome varies because of its rarity. Alison et al., estimated Gorlin syndrome at 1 per 40,000-60,000. In Asia prevalence of Gorlin syndrome is still uncertain. In some previous studies, the prevalence of Gorlin syndrome was reported to be 1 in 235,800 in Japan and 1 in 13,939,393 in Korea. A systematic review of Gorlin syndrome showed significant differences between ethnic groups. In East Asians, patients showed higher frequencies of cleft lips and palates, hypertelorism, and keratocystic odontogenic tumours. But in Northern Europe, patients showed significantly higher frequencies of BCCs, palmar and plantar pits, falx cerebri calcification, and family history. The disease affects both genders in rather equal distribution with the ratio of 1:1.3 in men and women respectively.

The mutation in patched 1 gene or known as PTCH1 (gene that encodes a transmembrane receptor that recognizes sonic hedgehog signaling proteins, a tumor suppressor gene located on chromosome 9q) has been associated with Basal cell nevus syndrome. It is known that 20% to 30% cases of BCNS represent De Novo mutations. The inactivation of the PTCH gene leads to tumorigenecity and the formation of multiple BCCs and other neoplasms.

Some features of Gorlin syndrome like BCC can present as early as infancy, with the median age of developing BCCs is in the 20s. The highlight feature of Gorlin syndrome is multiple BCCs. BCCs usually present as classic translucent papules with telangiectasias that may resemble...
acrochordons (skin tags). In 75% to 90% of patients, the superficial pits on the palms and soles can be found. Approximately 50% of cases reported epidermal inclusion cyst and milia. Odontogenic keratocyst presents with pain, swelling, and drainage of jaw cyst, maybe the initial reason for presentation and usually appears at a mean age of 13. The second most common malignancy found in Gorlin syndrome is medulloblastoma with 5% rate and occurs at a mean age of 2 and may experience mental retardation, seizures, and other neurologic abnormalities. Musculoskeletal abnormalities are very common with the rate of up to 75% and often found as a congenital condition. The musculoskeletal abnormalities can be bifid or splayed ribs (bifid rib), frontal bossing, cleft lip/palate, vertebral fusion, pectus excavatum, syndactyly, and hypoplastic thumbs. Hypertelorism, congenital blindness, strabismus, colobomas of retina or iris, and cataracts are some abnormalities that can be found in ophthalmologic abnormalities. Cardiac fibromas can cause bradycardia and usually happened during general anesthesia.9

Diagnosis of BCNS depends heavily on the criteria such as presence of two major clinical criteria or one major with two minor clinical criteria. Major criteria include: >2 BCCs or 1 BCC appears ≤ 20 years of age, histologic founding of odontogenic keratocysts of the jaw, palmar or plantar pitting, bilamellar calcification of the falk cerebri, bifid/ fused/ splayed ribs, first-degree relative with Gorlin syndrome. Minor criteria include: increased circumference of the head, medulloblastoma, cleft lip/palate, frontal bossing, moderate or severe hypertelorism, coarse facies, marked syndactyly of the digits, Sprengel deformity, pectus deformity, ovarian and cardiac fibromas, radiologic findings of hemivertebrae, bridging of the sella turcica, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet, or flame-shaped luencies of the hands or feet. Because the mutation of PTCH1 is the main cause of Gorlin syndrome, genetic testing for PTCH1 is suggested for the following conditions: (1) patients with family history but don’t meet the clinical criteria (predictive testing); (3) prenatal testing for patients with family history.9

Gorlin syndrome is a collection of neoplasms that needs a multidisciplinary approach and close surveillance to manage by each specialist in the organs involved. Adult patients should undergo complete skin checks for every 4 months while pediatric patients should get skin screening by a dermatologist annually until the first BCC is found, followed by screening at least every six months. Adults should have a complete skin check at least every four months.14,15 The most sensitive period for the onset of BCC is at the mean age of 25, therefore regular follow-ups are one of the best approaches that we can do, for us as physicians with limited resources.

The rarity of the syndrome is a challenge where there are no large clinical trials yet on how to best manage these patients by the time this report is being made. Our patient has congenital skeletal abnormalities but no other features related to Gorlin syndrome that were found in the age of 5 years old. However, adolescence and early adulthood is the most sensitive period with the median age of onset 25 years old for BCC. Therefore patients are required to do regular follow-ups and closely under observation of physicians. In this case, the patient was a boy of South Asian descent with a bifid rib on the 4th left rib. Although, since he is currently 5 years of age, there is a likely chance for him to develop tumour or cysts in the future if Gorlin syndrome are not ruled out by genetic testing.

CONCLUSION

Just like what is presented in this case, a patient with complaint of painless and solid chest lump with radiologic findings of bifid rib, Gorlin syndrome may be the first differential diagnosis for us physicians to work with. With barely any data on significance of bifid rib, and insignificant of genetic history, there is no conclusive exclusion for Gorlin Syndrome. We suggest close monitoring with regular follow-ups in such patients as the best approach for this condition. In addition, genetic testing for susceptible genes is strongly considered. However, in Indonesia, especially in Bali where cost and limited resources of genetic testing, observation and regular follow-up is the best management we can do as physicians, to detect the presence of any characteristic in Gorlin syndrome to ensure a better prognosis and quality of life for this patient.

CONFLICT OF INTEREST

The authors declare that, there is no conflict of interest.

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ETHICAL CLEARANCE

Patient or legal guardian had received signed written informed consent regarding publication of their respective medical data in medical journal with confidentiality aspect of personal information.

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AUTHOR CONTRIBUTION

All author had contributed to manuscript writing and agreed for the final version of manuscript for publication.

REFERENCE