CASE REPORT

Gorlin-Goltz syndrome: first reported case of bullae in the lungs complicated with tension pneumothorax

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SUMMARY

A 13-year-old girl was referred by her general practitioner with acute worsening exertional dyspnoea and sudden onset of left-sided chest pain. There was no associated trauma, palpitations or syncope. Clinical examination revealed that the left lung was hyper-resonant on percussion with reduced air entry on auscultation. Chest X-ray showed a left tension pneumothorax. She was treated conservatively with chest drain. Follow-up X-ray revealed multiple bullae within her left lung. Unfortunately, she redeveloped a pneumothorax and was sent to a tertiary centre. She was under the care of the paediatric cardiothoracic surgeons who organised a CT thorax and performed a lobectomy to remove the bullae. She was discharged from the tertiary centre and currently being followed up under the care of the paediatrician in the district general hospital. She have not developed any further pneumothoraces.

BACKGROUND

Gorlin-Goltz syndrome is a rare autosomal dominant multiorgan disorder; first described by Gorlin and Goltz in 1960.^{1 2} The classical triad of this syndrome are multiple basal cell carcinomas, odontogenic keratocyst and bifid ribs. However, there are more than 100 reported minor criteria in the literature. There are currently no described case of bullae within a Gorlin-Goltz syndrome patient after a literature search from Scopus, Web of Science and PubMed; we report the first case describing bullae in the lungs complicated with tension pneumothorax in a patient with Gorlin-Goltz syndrome.

CASE PRESENTATION

A 13-year-old girl was referred by her general practitioner due to worsening exertional dyspnoea. She have been complaining of intermittent breathlessness over the past 3 weeks; however, she developed a sudden onset of left-sided chest pain while walking her dog that prompted her to seek urgent medical attention. The chest pain was described as continuous stabbing and pleuritic in nature. The pain localises to the left side and worsen when she lies onto the left. She denies any history of coughing, trauma, palpitations, dizziness or syncope. She is a non-smoker, does not take any alcohol and has no history of recent foreign travel.

She has a medical history of Gorlin-Goltz syndrome diagnosed by a geneticist in a tertiary centre; where she has multiple mandibular cysts which was enucleated, pits in both palm and large head circumference.

Otherwise, she have been fit and well, with no regular medication or family history.

On examination, she was alert and talking in full sentences. The left lung was hyper-resonant on percussion and there was reduced air entry on auscultation. Respiratory rate was 28 per minute, heart rate of 98 beats per minute, saturating at 98% on air with a blood pressure of 136/76.

INVESTIGATIONS

Initial chest X-ray showed a left tension pneumothorax as shown in [figure 1](#). Follow-up chest X-ray after resolution of the pneumothorax revealed multiple bullae in her left lung as shown in [figure 2](#). CT thorax revealed a large bullae in the left lung as shown in [figure 3](#).

TREATMENT

A 12Fr chest drain was inserted using the Seldinger technique. Her pneumothorax was not resolving, hence the drain was placed on suction at 10cm/H₂O on day 3 and increased to 20cm/H₂O on day 4 of the chest drain insertion. The suction was taken off on day 6 and the chest drain was removed on day 7 when there was complete resolution of the pneumothorax. The subsequent chest X-ray revealed some bullae within the lungs.

Unfortunately, 2 days later she redeveloped a pneumothorax which was treated conservatively without a chest drain insertion. She was observed for further 2 days, in which she was transferred to a tertiary specialist centre for further management from the paediatric cardiothoracic surgeon as the pneumothorax was not resolving. She subsequently undergone a lobectomy to remove the bullae.

OUTCOME AND FOLLOW-UP

She was discharged from the tertiary centre and currently being followed up under the care of the paediatrician in the district general hospital. She have not developed any further pneumothoraces.

DISCUSSION

The Gorlin-Goltz syndrome is a rare autosomal dominant inherited syndrome manifested by multiple defects involving the skin, nervous system, eyes, endocrine system and bones. It is also known as basal-cell nevus syndrome, multiple basal-cell carcinoma syndrome, Gorlin syndrome, hereditary cutaneomandibular polyonocosis, multiple nevoid basal cell epithelioma-jaw cysts or bifid rib syndrome.¹

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Figure 1 Initial chest X-ray showing tension pneumothorax.

It was first reported by Jarisch and White in 1894 and later in detail by Gorlin and Goltz who described the classical triad of multiple basocellular epitheliomas, keratocysts in the jaws and bifid ribs.²

Epidemiology

Within literature, the prevalence of Gorlin-Goltz syndrome ranges between 1 in 57 000 to 1 in 256 000 inhabitants.^{3 4} This frequency vary geographically; for instance, in England, the frequency was 1 in 556 000 whereas, in Italy, it was 1 in 256 000 and Australia showed a prevalence of 1 in 164 000.⁴⁻⁶ The Gorlin-Goltz syndrome has equal predilection for both sex and present in all ethnic groups.^{7 8}

Genetics

Gorlin-Goltz syndrome is a disorder with high penetrance and variable phenotype expressiveness which can manifest itself spontaneously.⁸ Individuals affected by the Gorlin-Goltz

Figure 2 Chest X-ray showing bullae within the left lung.

Figure 3 CT showing bullae within left lung.

syndrome show very different signs and symptoms despite having similar molecular alteration.⁸ It has been suggested that there is a complex variability of phenotype due to the interaction between genetic and environmental factors.

It is considered that between 30% and 50% of patients who suffer from this syndrome do not know if any of their family members has had it.⁸ The mutation of the 'Patched' (PTCH) tumour suppressor gene has been identified to cause the Gorlin-Goltz syndrome which is located at chromosome 9q22.3. This gene codes a transmembrane glucoprotein that is found in the Hedgehog signalling pathway which regulates multiple processes such as embryogenesis, homeostasis maintenance in old tissue, tissue repairing during persistent chronic inflammation and carcinogenesis.⁸

Clinical pathological aspect

Diagnosis of Gorlin-Goltz syndrome can be established when two major or one major and two minor criteria are present.^{1 9} In 1993, Evans *et al* described diagnosis made by the most frequent and specific features of the syndrome.⁴

The major criteria are multiple basal-cell carcinoma or one occurring under the age of 20 years; odontogenic keratocysts; palmar or plantar pits; calcification of the falx cerebri; bifid, fused or markedly splayed ribs and first-degree relative with nevoid basal-cell carcinoma syndrome.^{1 8 10}

While minor criteria are macrocephaly determined after adjustment for height; congenital malformations: cleft lip or palate, frontal bossing, 'coarse face', moderate or severe hypertelorism; skeletal abnormalities: Sprengel's deformity, marked pectus deformity, marked syndactyly of the digits; radiological abnormalities: bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modelling defects of the hands and feet or flame-shaped lucencies of the hands or feet; ovarian fibroma and medulloblastoma.^{1 8 10} Amezcaga *et al* summarised more than 100 minor features described in the literature as shown in table 1.

We describe the first case of bullae within the lungs of a patient with Gorlin-Goltz syndrome. Due to lack of recognition, patient has suffered a serious complication of tension pneumothorax.

Diagnosis and management

Early diagnosis is important to reduce the complications of the symptoms such as malignant tumours and destruction deformities of the oromaxillofacial region.⁸ Since the Gorlin-Goltz

Table 1 Anomalies described in Gorlin-Goltz syndrome^{1 8 10}

Skeletal anomalies	
Bridging sella turcica	Syndactyly/oligodactyly
Scoliosis	Shortened fourth metacarpal
Bifid rib	Splayed/fused rib
Occult bifid rib	Cervical rib
Absent rib	Sprengel scapular deformity
Hemivertebrae	Pelvic calcification
Flatfoot	Pectum excavatum/carinatum
Polydactyly	Arachnodactyly, hallux valgus
Skin anomalies	
Basal cell carcinoma	Milia
Palmar/plantar pits	Comedones
Craniofacial anomalies	
Calcification of the cerebral falx	Brachycephaly
Macrocephaly	Tentorium cerebellum calcification
Frontal bossing	Bridged sella turcica
Coarse face	Coroidal cyst
Sexual anomalies	
Uterine/ovarian fibromas	Supernumerary nipple
Ovarian fibrosarcoma	Hypogonadism/cryptorchidism
Calcified ovarian cysts	Female distribution of pubic hair
Gynaecomastia	
Neurological anomalies	
Medulloblastoma	Agenesis/disgenesis of corpus callosum
Meningioma	Schizophrenic personality
Mental retardation	Nervous deafness
Congenital hydrocephalus	Anosmia
Ophthalmic anomalies	
Hypertelorism	Glaucoma
Exotropia	Choroidal/optic nerve coloboma
Congenital amaurosis	Congenital blindness and opaque cornea
Ptosis	Cataracts
Internal strabismus (convergent/divergent)	Chalazion
Orofacial anomalies	
Odontogenic keratocysts	Palate or maxillary sinus fibroma
High-arch palate/prominent palatine ridges	Malocclusion (maxillary hypoplasia, mandibular hyperplasia, cleft palate)
Cleft lip/palate	Fibrosarcoma of the jaws
Impacted teeth/agenesis	Ameloblastoma
Other	
Inguinal hernia	Renal anomalies
Lymphomesenteric cysts	Left ventricular fibroma
Cardiac fibroma	

syndrome affects multiorgan, a multidisciplinary management is highly recommended, which includes oromaxillofacial surgeons, oral pathologists, paediatricians, dermatologists and geneticist in the diagnosis, treatment and follow-up of these patients.⁹

It is also crucial to involve families in regular screening as it may be possible to prove whether someone in a family has inherited the condition or not by DNA tests, either by tracking the chromosome 9 containing the faulty gene or by direct analysis of the mutation causing the disease in a particular family.⁹

Patient should have an orthopantomography, frontal-back-thorax radiograph and craniofacial CT or MR to identify osseous morphology. In addition, female patients should be offered a pelvic ultrasound.⁸ All patient should be followed up yearly to monitor and treat symptoms early to prevent secondary complications. The treatment to Gorlin-Goltz syndrome is specific to the clinical manifestations and there is currently no cure to the syndrome.

Learning points

- ▶ Patient with Gorlin-Goltz syndrome can develop bullae within the lungs.
- ▶ Ruptured lung bullae can cause a pneumothorax and if went unrecognised, it will progress to a tension pneumothorax.
- ▶ At present, there is no treatment towards Gorlin-Goltz syndrome but early recognition can prevent secondary complications.

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