Radiology Case Reports 14 (2019) 448–451

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Case Report

Radiography of Chitayat syndrome in an infant male

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ARTICLE INFO

Article history:
Received 12 October 2018
Revised 13 January 2019
Accepted 13 January 2019
Available online 24 January 2019

Keywords:
Chitayat
Genetic
Syndrome
ERF
Gene
Bronchomalacia

ABSTRACT

Chitayat syndrome is a rare genetic syndrome characterised by bilateral hyperphalangism, bronchomalacia, hallux valgus, and other facial dysmorphism including large anterior fontanelle, hypertelorism, and anteverted nostrils. Since the initial discovery, only few cases of Chitayat syndrome have been reported in the literature. Previous literatures showed the genetic link between 5 case reports, showing that a unique link of recurrent c.266A>G p.(Tyr89Cys) variant in the ERF gene may be the contributory genetic cause of Chitayat syndrome. However, it still remains as an unfamiliar genetic syndrome. In this case report, we aim to discuss a rare case of Chitayat syndrome and demonstrate the radiological findings associated.

Case report

This patient was the third child of nonconsanguineous healthy parents of Bangladeshi origin. His 2 siblings are unaffected and there is no other contributory family history. He was born at 39 weeks' gestation with birth weight of 3340 g (25th centile) and head circumference of 34.5 cm (25-50th centile). The delivery was spontaneous and uncomplicated with APGAR scores of 9 at 1 minute and 10 at 5 minutes. On examination at birth, dysmorphic features were noticed. These included low posterior hairline, epicanthic folds, flat nasal bridge, bilateral medially deviated brachydactyly of index fingers, clinodactyly of ring and little fingers (Fig. 1) and bilateral hallux valgus (Fig. 2).

His early postnatal period was complicated by progressive tachypnoea at around 6 hours of life, requiring oxygen therapy and leading to Neonatal Intensive Care Unit admission. He was treated as respiratory distress of unknown cause. Oxygen supplement was given via nasal cannula. By day 11, he was

Competing Interests: The authors have declared that no competing interests exist.

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https://doi.org/10.1016/j.radcr.2019.01.003

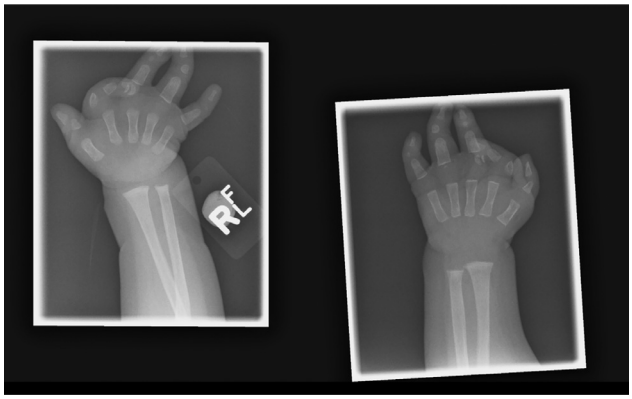

Fig. 1 – Plain radiograph of patient's right hand showing bilateral medially deviated brachydactyly of index finger and clinodactyly of ring and little fingers. The proximal phalanges appear hypoplastic and delta shaped, demonstrating typical phenotype in Chitayat's Syndrome.

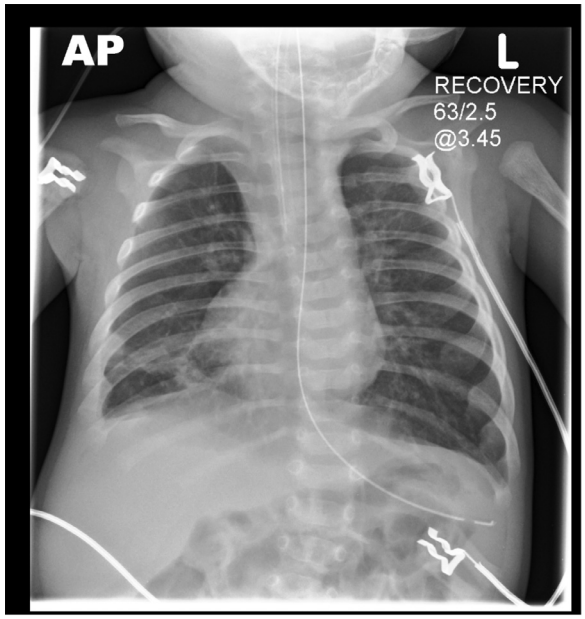

Fig. 3 – Plain chest radiograph showing hyperinflation of both lungs, atelectasis of right lower lobe with bilateral perihilar peribronchial thickening, likely related to viral bronchiolitis. Endotracheal tube in place. The foetal heart is normal situs.

breathing room air and his tachypnoea had resolved by day 22 of life. He was discharged home with a stable respiratory rate on room air and bottle-fed.

In the first few months of life he required further 2 separate admissions for respiratory distress secondary to bronchiolitis (Fig. 3). The first admission at 8 weeks of age was managed with high frequency oscillation ventilation. Following this admission, he required feeding via a nasogastric tube due to ongoing difficulties with tachypnoea during bottle feeds. The second episode occurred at 12 weeks of age, but was less severe and managed conservatively with antibiotics alone. His respiratory difficulties were considered to be secondary to bronchomalacia, requiring home oxygen therapy until the age of 11 months. The diagnosis of bronchomalacia was confirmed with a bronchoscopy at the Great Ormond Street Hospital. The patient's weight in the first year of life remained in 3rd centile (Fig. 4), reflecting his feeding difficulties secondary to respiratory symptoms.

A sample was sent to the Great Ormond Street Hospital in London at 20 weeks of age for genetic analysis. This revealed a variant in ERF: c.266A>G p.(Tyr89Cys) consistent with a diagnosis of Chitayat syndrome.

Discussion

Chitayat syndrome is a rare genetic syndrome associated with hyperphalangism, facial abnormalities, and diffuse bronchomalacia, first described in 1931 [1]. Balasubramian et al. described the common phenotypic features found in Chitayat syndrome including respiratory compromise, bilateral accessory phalanx, clinodactyly, hallux valgus, and facial features of

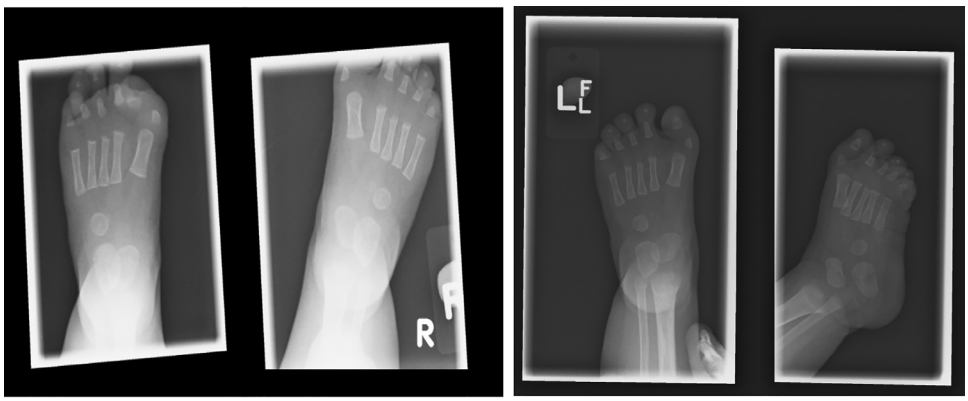

Fig. 2 – Plain radiograph of patient's left and right foot showing hallux valgus.

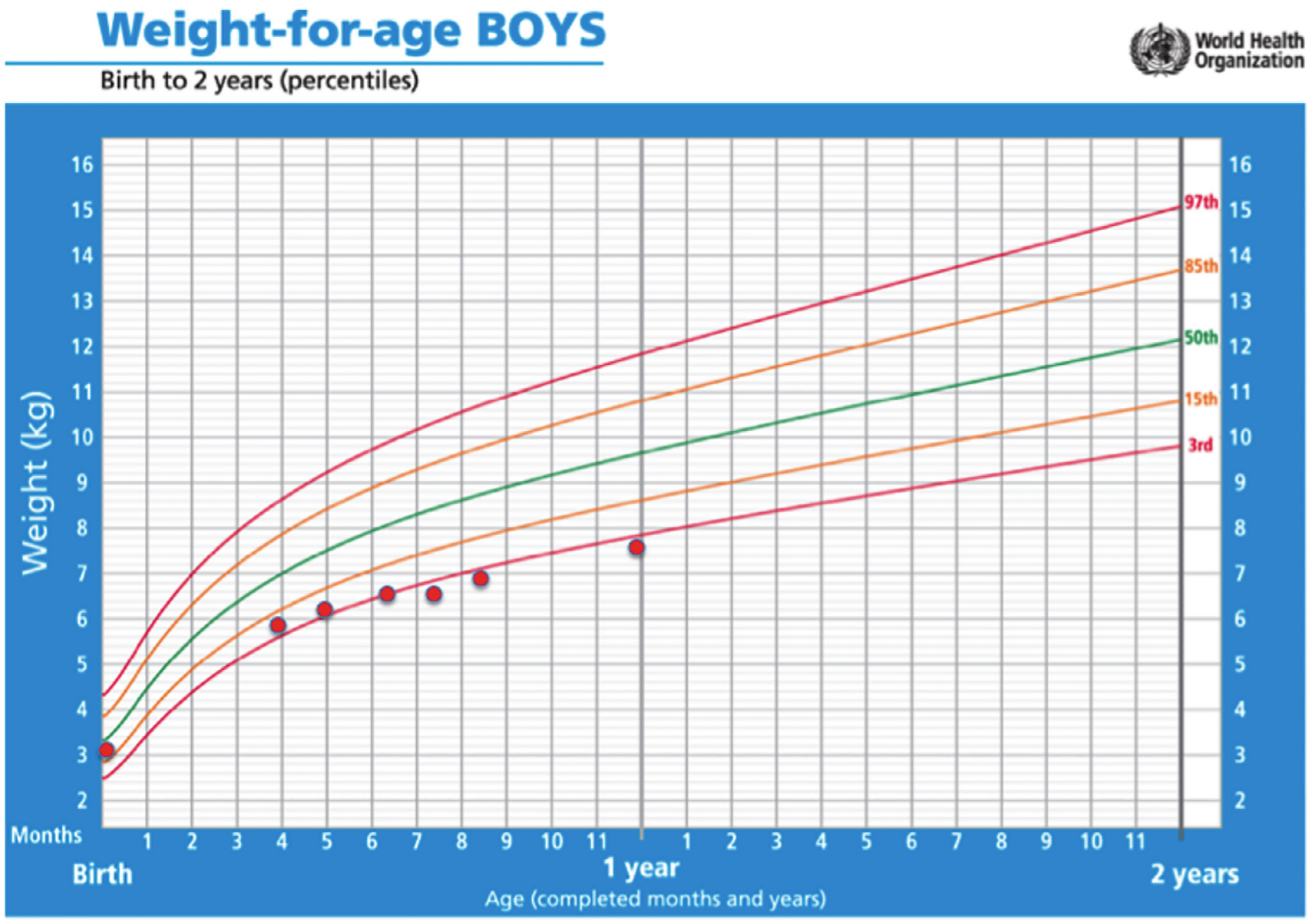

Fig. 4 – Growth chart of patient's weight illustrated on WHO official chart.

large anterior fontanelle, prominent eyes, hypertelorism and depressed nasal bridge [2]. Their case report identified 5 individuals with same phenotypic findings. Genetic analysis of all 5 cases discovered a recurrent, novel variant NM_006494.2: c.266A>G p.(Tyr89Cys) in ERF gene.

Our case study demonstrates similar phenotypic features as described in the literature. These features include low posterior hairline, flat nasal bridge, bilateral medially deviated brachydactyly of index fingers, clinodactyly of ring and little fingers and bilateral hallux valgus. The proximal phalanges appear hypoplastic and delta shaped, likely contributing to the deviation.

Our patient was hospitalised with respiratory distress requiring intensive care input immediately after birth. This correlates to the initial findings described by Chitayat et al. of respiratory distress presenting at birth [1]. He required multiple hospital admissions for respiratory distress of unknown cause and one occasion of bronchiolitis. Similar episodes of respiratory distress have been reported by Balasubramian et al. The underlying diagnosis of bronchomalacia was identified in all 5 patients [2]. In our case, his frequent requirement of respiratory support led to further investigations including bronchoscopy which revealed bronchomalacia.

In view of bilateral brachydactyly of index fingers of our patient, a diagnosis of Catel-Manzke syndrome was considered. Catel-Manzke syndrome was first described by Catel in 1961 as a palatodigital syndrome with Pierre Robin anomaly [3]. Phenotypic facial features include micrognathia and glossoptosis, hyperphalangism, and brachydactyly of index fingers. Manzke et al. [4] described a spectrum of Catel-Manzke's syndrome with some patients showing isolated anomaly without Pierre Robin sequence, consisting of small narrow nostrils and posteriorly set ears. Mutations in the TGDS gene has been reported to be the contributory factor [5]. In our case, the distinct dysmorphic facial features did not correlate with Catel-Manzke syndrome.

Temtamy preaxial brachydactyly hyperphalangism syndrome (TPBS) is another condition considered as a diagnosis. TPBS is associated with brachydactyly, hyperphalangism, craniosynostosis, micrognathia, and deafness. In 2013, Low et al. described pectus excavatum as a novel phenotype of TPBS [6]. Genetic basis of TPBS is associated with mutations of the CHSY1 gene [7]. The lack of hearing defects or pectus excavatum with our patient prompted for other diagnosis.

Genetic analysis of our patient was performed at the Great Ormond Street Hospital in London at 20 weeks of age. This

revealed a variant of c.266A>G p.(Tyr89Cys) in ERF gene, which was consistent with the genetic analysis findings demonstrated in Balasubramian et al. [2] confirming the diagnosis of Chitayat Syndrome.

We have described a rare case of Chitayat syndrome. This is an additional study for delineation of genetic findings established in 2017 for Chitayat syndrome which is an unusual yet important cause of respiratory compromise for newborns to be aware of.

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