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Congenital Hallux Varus: A Rare Prenatal Detection and Postnatal Management

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Abstract

Congenital hallux varus (CHV) is an uncommon anomaly of the forefoot where the big toe is positioned medially at the metatarsophalangeal joint, accompanied by a significant gap between the first and second toes. CHV is associated with difficulty in wearing clothes and footwear. CHV can cause pain that hampers the quality of life and can often be aesthetically displeasing.

Although prenatal presentation is rare, CHV can be detected by antenatal ultrasound with appropriate foot examination and a high index of suspicion. Early detection and management are recommended for optimizing antenatal and postnatal management as they may reduce the chance of permanent deformity.

We are herewith reporting a prenatally detected case of a CHV with polysyndactyly and its management. This is one of the few prenatally reported cases of isolated unilateral foot abnormalities.

Keywords

- congenital hallux varus
- surgical treatment
- medial deviation
- congenital
- great toe

Introduction

Congenital hallux varus (CHV) is a rare anomaly of the forefoot where the great toe is positioned medially at the metatarsophalangeal joint, accompanied by a significant gap between the first and second toes. Hallux varus is a very rare deformity as compared to hallux valgus and the congenital type is uncommon.[1]

This is associated with difficulty in wearing clothes, and footwear, even more pain that hampers the quality of life and is cosmetically unacceptable.[1] Failure of early diagnosis or insufficient correction may result in soft-tissue imbalance leading to osseous changes, and persistent deformity due to ongoing soft-tissue contractures. The present case report describes a clinical presentation and management of a prenatally detected case of CHV with polysyndactyly.

Case Report

A 31-year-old primigravida booked at 10 weeks. She had conceived spontaneously in a consanguineous marriage. Her blood group was Rh negative, while the father's blood group was Rh positive. The indirect Coombs test was negative. She had no history of medical illness or prolonged use of any medications. There was no significant family history.

Ultrasound (US) was performed on a GE Voluson S8 unit with a C 1–5 curvilinear probe. A nuchal translucency scan at 12 weeks was reported as normal and combined screening was

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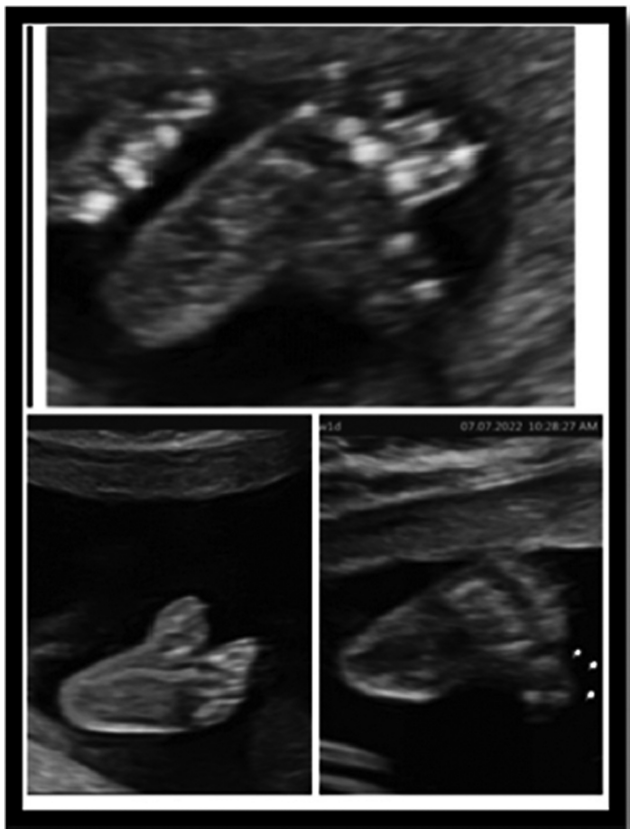


Fig. 1 Two-dimensional ultrasonography examination suggestive of congenital hallux varus with polysyndactyly.

low risk for common aneuploidies. An anomaly scan at 19 weeks showed normal fetal growth and amniotic fluid. The fetal left foot was found to show a medially angled great toe at the metatarsophalangeal joint, almost at a 90-degree angle.

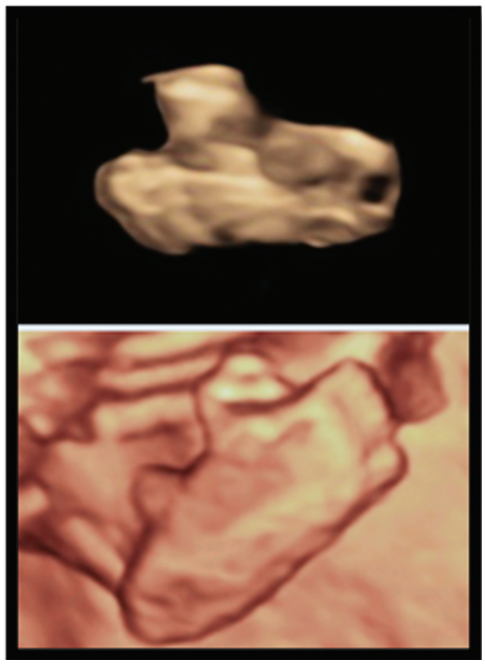
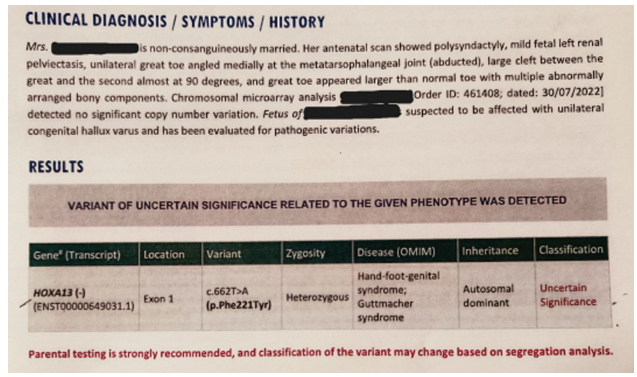


Fig. 2 Three-dimensional rendered image of foot.



CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Mrs. [redacted] is non-consanguineously married. Her antenatal scan showed polysyndactyly, mild fetal left renal pelviectasis, unilateral great toe angled medially at the metatarsophalangeal joint (abducted), large cleft between the great and the second almost at 90 degrees, and great toe appeared larger than normal toe with multiple abnormally arranged bony components. Chromosomal microarray analysis [redacted] Order ID: 461408; dated: 30/07/2022] detected no significant copy number variation. *Fetus of* [redacted] suspected to be affected with unilateral congenital hallux varus and has been evaluated for pathogenic variations.

RESULTS

VARIANT OF UNCERTAIN SIGNIFICANCE RELATED TO THE GIVEN PHENOTYPE WAS DETECTED

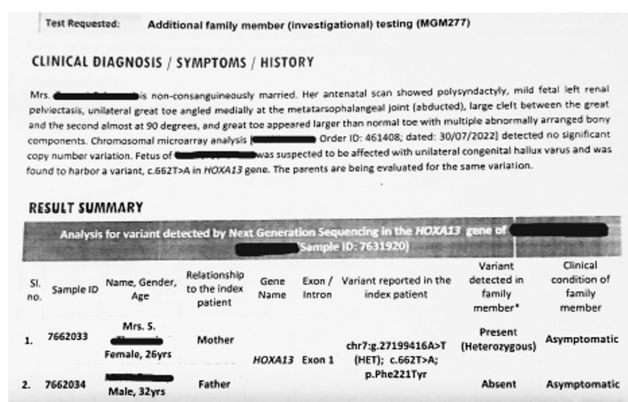
Gene* (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
HOXA13 (-) (ENST00000649031.1)	Exon 1	c.662T>A (p.Phe221Tyr)	Heterozygous	Hand-foot-genital syndrome; Guttmacher syndrome	Autosomal dominant	Uncertain Significance

Parental testing is strongly recommended, and classification of the variant may change based on segregation analysis.

Fig. 3 The fetal whole exome sequence report.

There was a large cleft between the great and second toes. The great toe was abnormally broad, with nonlinear arrangements of multiple phalangeal bones within (►**Fig. 1**). The three-dimensional-rendered images showed similar findings (►**Fig. 2**). The contralateral lower limb and both upper limbs were normal. Fetal renal pelvis measurements were 4.5 and 5 mm, indicating mild bilateral renal pelviectasis. Renal pelvis measurement of more than 4 mm is pelviectasis for this gestation. No other systemic abnormalities were noticed on US.

A diagnosis of unilateral CHV with polysyndactyly and bilateral mild renal pelviectasis was made. Because of the structural abnormality, the presence of a minor soft marker and a history of consanguinity, amniocentesis was offered to rule out an underlying genetic causes. An injection of anti-D prophylaxis was administered to avoid isoimmunization. An uneventful amniocentesis was performed and the sample was sent for chromosomal microarray (CMA) and whole exome sequencing (WES). CMA showed no copy number variations. WES revealed a heterozygous missense variant (C.662T > A) in exon 1 of the HOX A 13 gene, which was reported as a variant of unknown significance with autosomal dominant inheritance for hand-foot-genital syndrome and Guttmacher syndrome (►**Fig. 3**). This mutation usually presents severe limb abnormalities and genital abnormalities but can be seen in healthy individuals too. Couple carrier testing showed that mother carries a similar variant of HOX A 13 at EXON 1 in a heterozygous and is an asymptomatic carrier (►**Fig. 4**). Phenotype and



Test Requested: Additional family member (investigational) testing (MGM277)

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Mrs. [redacted] is non-consanguineously married. Her antenatal scan showed polysyndactyly, mild fetal left renal pelviectasis, unilateral great toe angled medially at the metatarsophalangeal joint (abducted), large cleft between the great and the second almost at 90 degrees, and great toe appeared larger than normal toe with multiple abnormally arranged bony components. Chromosomal microarray analysis [redacted] Order ID: 461408; dated: 30/07/2022] detected no significant copy number variation. Fetus of [redacted] was suspected to be affected with unilateral congenital hallux varus and was found to harbor a variant, c.662T>A in *HOXA13* gene. The parents are being evaluated for the same variation.

RESULT SUMMARY

Analysis for variant detected by Next Generation Sequencing in the *HOXA13* gene of [redacted] Sample ID: 7631920)

Sl. no.	Sample ID	Name, Gender, Age	Relationship to the index patient	Gene Name	Exon / Intron	Variant reported in the index patient	Variant detected in family member*	Clinical condition of family member
1.	7662033	Mrs. S. [redacted] Female, 26yrs	Mother	*HOXA13*	Exon 1	chr7:g.27199416A>T (HET); c.662T>A; p.Phe221Tyr	Present (Heterozygous)	Asymptomatic
2.	7662034	[redacted] Male, 32yrs	Father				Absent	Asymptomatic

Fig. 4 Maternal carrier testing.

Fig. 5 Picture of the neonatal foot suggestive of congenital hallux varus with polysyndactyly.

genotype did not correlate. Detailed genetic counseling by a geneticist helped the couple in further decision-making. The couple decided to continue the pregnancy.

A detailed pediatric orthopedic consultation was done to explain the implications of this condition and the required postnatal surgery. The Rest of the antenatal period was uneventful, and serial US examinations showed normal with normal growth, normal Doppler and normal affected limb movement. She delivered a male baby weighing 2.8 kg at 39+ weeks vaginally. APGARs were normal. Postnatal examination findings were consistent with the prenatal findings. The left foot had a medially placed broad great toe with a single nail, almost at 90 degrees. There was evidence of rudimentary skin and soft-tissue elements with multiple creases on them indicating polysyndactyly. Rest of the toes were normal (►**Fig. 5**). Hands and genitalia were normal. A neonatal US abdomen showed normal kidneys. The pelviectasis resolved.

Postnatal X-ray findings were concordant with antenatal US, which showed a broad great toe angled medially with small multiple phalanges with the irregular arrangement in the great toe and the presence of excess soft-tissue elements in the preaxial region suggestive of polysyndactyly (►**Fig. 6**).

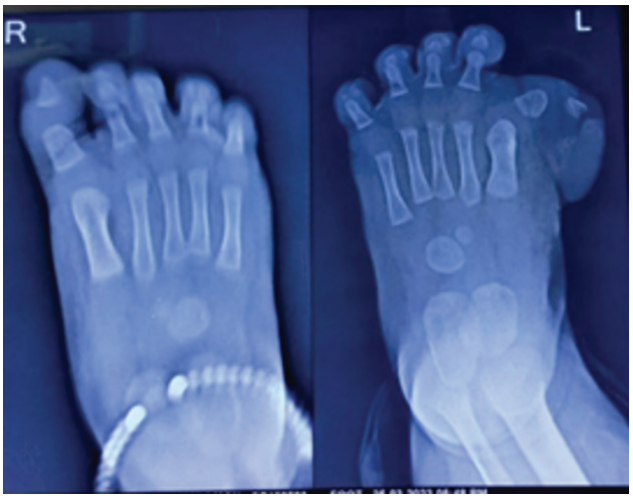


Fig. 6 X-ray showed small multiple phalanges with rudimentary soft-tissue elements.

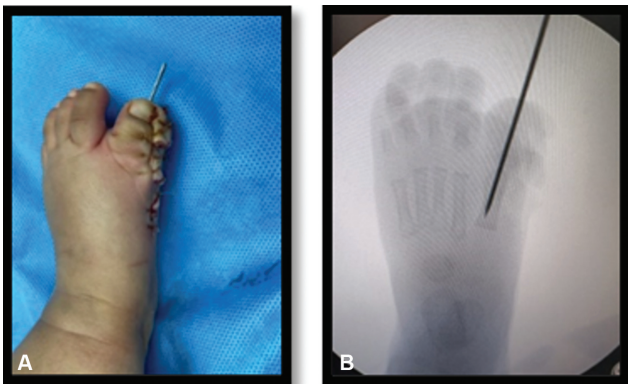


Fig. 7 (A, B) Postsurgical picture: Excision, Z-plasty with Kirschner wire fixation.

At the age of 5 months, an infant's surgery was performed. Rudimentary segments of bones, skin, and soft tissue were excised and a Z-plasty followed by a Kirschner wire (K-wire) insertion was performed by a pediatric orthopedic surgeon with an overall optimized correction (►**Fig. 7**). K-wire was removed on the 20th day postsurgery. Wound healing was optimal. The recurrence of the deformity and the possible need for future surgery were explained to the parents. At the follow-up visit, after 3 months postsurgery, the great toe appeared unified with a normal angle and the space between the first and second toe normalized (►**Fig. 8**).

Discussion

CHV known as an atavistic great toe is a rare forefoot abnormality in which the great toe is angled medially at the metatarsophalangeal joint and there is the presence of a large cleft

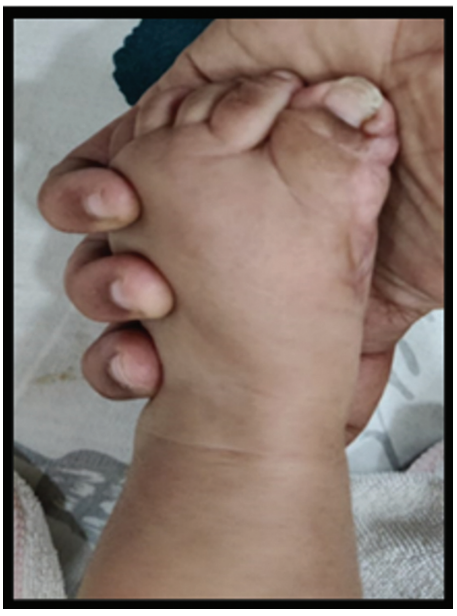


Fig. 8 Infant foot picture at 3 months postsurgery.

between the first and second toe. This deformity is characterized by a medial deviation of the big toe and an enlarged web space, or the presence of a cleft between the first two toes.[2,3]

Different etiological factors have been hypothesized by various researchers for the development of Hallux varus. This can be congenital as well as acquired. CHV is less commonly noted than the acquired type and is commonly diagnosed in infancy. It usually presents once a child starts walking or in adulthood. It presents as a varus deformity of the first toe or can be incidentally found on clinical examination and X-rays in the case of milder forms. Very rarely, this entity may present during the prenatal period.[4]

CHV has various etiologies like thickened medial cords, the first metatarsal longitudinal epiphyseal bracket, slopes, short blocks of short first metatarsals, and ineffective abductor hallucis and adductor hallucis insertions.[3] Farmer reported that the cause of isolated CHV could be a duplication of the first ray of digits, but it is not a real duplication, hence the toe appears wide and broad.[2] Sometimes it may be associated with other limb abnormalities and polydactyly. In this case, the cause for CHV was a taut fibrous band running from the medial side of the great toe to the base of the first metatarsal, which has created medial deviation with longitudinal bracket epiphysis of the first metatarsal with preaxial polydactyly and small-sized phalanges.

The exact genetic association and inheritance pattern of isolated cases of CHV are not found in the various reported cases. CHV does have an association with connective tissue disorders with varied inheritance. One of the cases by Sebahat Atar Gurel et al found a coincidental association with the pericentric inversion of chromosome 9[inv (9)], one of the most common structurally balanced chromosomal variations. Following a literature search, the exact genetic etiopathogenesis was not established. Because inv (9) is commonly seen in the normal population, this was a coincidental finding as per this case report.[5]

In our case, a variant of unknown significance was detected that was a heterozygous missense variant at exon 1 of the HOX A 13, which has an autosomal dominant inheritance for hand-foot-genital syndrome. Hand-foot-genital syndrome and Guttmacher syndrome affect the distal limbs (minor manifestations) and genitourinary tract. In a case study by Goodman et al, a nonsense mutation in the homeobox of HOXA13 has been identified in one affected family. They have found that mutations in HOXA13 can cause more severe limb abnormalities and genital abnormalities.[6] But in the present case, the phenotype did not exactly match with the mentioned variant. Fetal genitalia appeared normal (as per the PCPNDT act we did not disclose the sex of the baby). As per the Genomad database, this variant was found in healthy individuals. Considering the possibility of a mild manifestation of the disease and the fact that this condition was not associated with an intellectual disability or any other major disability except that it needed postnatal surgery and cosmetic corrections, the couple decided to continue the pregnancy till the term after thorough counseling.

If the deformity is not addressed at the earliest or is undercorrected, the soft-tissue imbalance creates osseous changes and soft-tissue contractures lead to persistent deformity in adulthood.[7] Patients experience pain and difficulty with footwear if this condition is left untreated. This condition may recur even after the operative procedure secondary to under-corrections or recurrence.

It can be managed conservatively or surgically. To date, various surgical techniques have been proposed for the correction of CHV like abductor hallucis muscle release for mild and resistant deformities, excision of the epiphyseal bracket if the epiphyseal bracket is found to be the cause of CHV, and the Farmer technique for moderate-to-severe deformities. These techniques can be divided into soft-tissue procedure, bony procedure, or a combination of both soft tissue and bony procedures. The most common soft-tissue procedures described in the literature are by McElvenny and Farmer. McElvenny proposed the removal of accessory bones, sesamoidectomy, capsulotomy, and medial fibrous band release, followed by transfixing the metatarsophalangeal joint with a K-wire.[8] Farmer described a rotational skin flap and syndactylisation involving the first and second toes.[2]

A combined technique of soft tissue and bony procedures is the preferred method for optimum surgical success. Correction of the hallux varus angle and widening of the first web space are essential for good clinical and cosmetic outcomes.[9] In the present case, the pediatric orthopedic surgeon who had operated on the baby performed an excision of excess rudimentary bones and soft tissue and the release of the fibrous band with K-wire fixation and Z-plasty. The procedure involved both soft tissue and bony components.

Postsurgery follow-up after 3 months showed a well-healed, well-aligned great toe and cosmetically acceptable foot anatomy (►**Fig. 8**). Parents were counseled about the risk of recurrence and advised to wear opposite-side footwear to avoid recurrence. Counseling reiterated the need for long-term followup to identify any recurrence that may require repeat surgery.

Conclusion

Incorporation of the foot in routine US examinations helps in diagnosing isolated foot abnormalities.

Though this is not a lethal condition, it does have a bearing on the quality of life. Early detection and early surgical management are the keys to satisfactory outcomes.

Genetic association has a greater implication. Hence, genetic evaluation is a must, as was done in our case. Multidisciplinary team management and detailed genetic counseling are necessary in such cases before taking decisions.

This case report of a rare congenital form of CHV, which was detected in the prenatal period, raises awareness about the rare condition.

Conflict of Interest
None declared.

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