Case Report

Isolated Ectrodactyly in a Late Preterm Neonate: A Case Report

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ABSTRACT

Ectrodactyly, also known as Split Hand Foot Malformation (SHFM), is a rare congenital limb defect characterized by the partial or complete absence of the central rays of the hands and feet, resulting in a varied spectrum of anomalies ranging from mild digital deficiencies to the classic "split" appearance of the affected limb. It may involve a single or multiple extremities, and its presentation can vary widely even within the same family. Ectrodactyly may occur as an isolated malformation or as part of a broader syndrome, often associated with additional craniofacial, auditory, or neurodevelopmental abnormalities. Although the exact etiology is heterogeneous, genetic factors play a significant role, with several chromosomal regions and genetic mutations implicated in their pathogenesis. Early recognition of SHFM is important for timely evaluation, genetic counselling, planning functional and reconstructive interventions, and rehabilitation. In this report, we present a case of SHFM, outlining its clinical presentation and discussing the diagnostic and therapeutic considerations relevant to its management.

Key words: Ectrodactyly, Foot malformation, Congenital.

Ectrodactyly, also known as "Lobster Claw Hand" or "Split Hand or Foot Malformation (SHFM)", is a rare congenital malformation of the extremities affecting 1 in 90,000 babies with no sex predilection [1, 2]. First described by Theodor Kerckring, a 17th-century Dutch anatomist. It involves the central digits of the feet or hands, syndactyly, and a median cleft of the phalanges, metacarpals, or metatarsals, resulting in a characteristic lobster-claw appearance of the hands or feet [3, 4]. The basic embryologic abnormality involves failure to maintain the normal functioning of the apical ectodermal ridge (AER), leading to impaired differentiation of the autopod (hand or foot). Limb development is regulated by interactions between three specialized cell groups- AER, the progress zone (PZ), and the zone of polarizing activity (ZPA).

The AER plays a key role by promoting mesenchymal cell proliferation in the PZ, directing proximodistal limb development. Disruption or environmental damage to the AER can lead to ectrodactyly [5, 6]. It can occur in two forms: isolated or as part of a syndrome. Both forms may be sporadic or familial, with sporadic cases being more common. In familial cases, the most common mode of inheritance is autosomal dominant with variable expressivity (associated with SHFM Types 1, 3, 4, and 5), though autosomal recessive (SHFM Type 6- homozygous mutation in WNT10B gene) and X-linked (SHFM Type 2) patterns have also been reported rarely. Syndromic associations include EEC syndrome (ectrodactyly, ectodermal dysplasia, cleft lip/palate syndrome), ADULT syndrome (acro-dermato-ungual-lacrimal-tooth syndrome), CHARGE syndrome (coloboma of the eye, heart defects, atresia of nasal choanae, retardation of growth/development, genito-urinary anomalies, ear anomalies), LADD syndrome (lacrimo-auriculo-dento-digital syndrome) or VACTERL association (vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies and limb anomalies) [5, 6]. A case of a late preterm neonate with isolated ectrodactyly of bilateral lower limbs with an uneventful antenatal and postnatal course is presented here, outlining its clinical presentation and discussing the diagnostic and therapeutic considerations relevant to its management.

CASE PRESENTATION

A 29-year-old mother gave birth to a male neonate at 36+3 weeks of gestation via spontaneous vaginal delivery after 4 years of second-degree consanguineous marriage. The mother has been pregnant three times (gravida 3), with no prior pregnancies reaching the age of viability (para 0), no living

Access this article online

Received – 18th February 2026
Initial Review – 25th February 2026
Accepted – 01st April 2026

DOI: 10.32677/ijch.v13i4.8126

Quick Response Code

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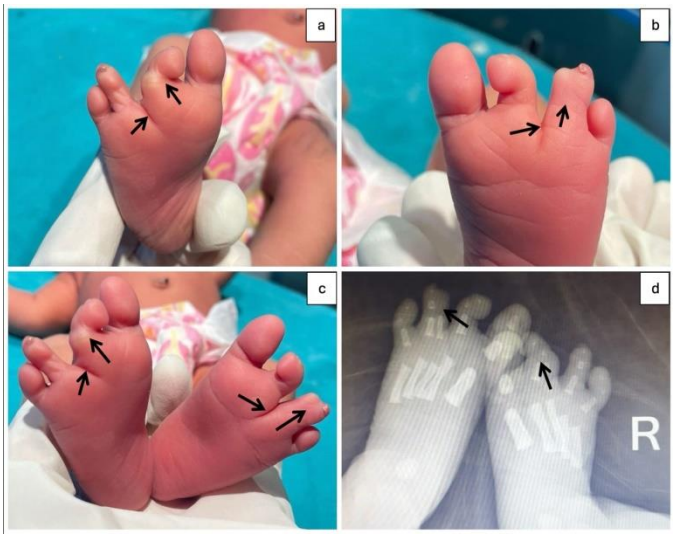
children (living 0), and a history of two pregnancy losses before viability (abortions 2): G3P0L0A2. The antenatal period was unremarkable without any exposure to a teratogenic drug or radiation.

Antenatal anomaly scan did not reveal any limb defects. The neonate was late preterm and had a birth weight of 2610 grams, which was appropriate for gestational age as per Intergrowth-21 charts. The neonate cried immediately after birth, and delayed cord clamping was done. The neonate was then put on early skin-to-skin contact when he was noted to have a deep cleft of both feet with syndactyly of the 2nd and 3rd toes of the right foot and 3rd and 4th toes of the left foot (Figure 1a, b, c).

The rest of the general and systemic examination was normal. Infantogram showed absent middle and distal phalanx of the 2nd toe of the right foot and 3rd toe of the left foot with juxtaposed proximal phalanges of the affected toes (Figure 1d). Figure 1. Ectrodactyly of bilateral feet. (a, b, c) Black arrows show a deep cleft of both feet with syndactyly of the 2nd and 3rd toes of the right foot and the 3rd and 4th toes of the left foot. (d) X-ray shows absent middle and distal phalanx of the 2nd toe of the right foot and 3rd toe of the left foot with juxtaposed proximal phalanges of the affected toes.



Routine pulse oximetry screening, hearing screening, and cord TSH were normal. Eye examination, ultrasound abdomen, hearing screening, and echocardiography (ECG) were also normal. On detailed history, a similar foot deformity was also present in the paternal uncle. Considering the positive family history and typical findings, a clinical diagnosis of SHFM was made. An orthopedic team opinion was sought, and corrective surgery was planned for 1-2 years of age. Parents were advised to do genetic testing, but refused due to financial issues. Postnatal course was uneventful, and the neonate was discharged on day 3 of life on exclusive breastfeeding. On the latest follow-up at 12 months of age, he had appropriate age-development milestones and could stand without support and walk with support. The child is currently under orthopedic follow-up and is planned for surgery.

DISCUSSION

Ectrodactyly involves the central digits of the hands or feet and is characterized by syndactyly with a median cleft of the extremity, producing the typical “lobster-claw” appearance. The present case had an unremarkable antenatal history but was noted to have a deep cleft with syndactyly affecting the toes of both feet at birth. A similar limb defect was present in a second-degree relative. The neonate had an uneventful postnatal course and was discharged on day 3 of life. Prenatal exposure to certain medications, such as acetazolamide, has been associated with an increased risk of isolated ectrodactyly [7]; however, no history of teratogenic drug exposure was present in this case. Ectrodactyly can be detected on antenatal ultrasonography, with three-dimensional ultrasonography being more sensitive than conventional gray-scale imaging for identifying limb anomalies [5].

Several cases of antenatally detected ectrodactyly have been reported in the literature. Pinette et al. described bilateral ectrodactyly of the hands and feet identified on level II obstetric ultrasound in a primigravida mother [1]. Durmaz et al. reported another case of a 22-year-old woman with an antenatally detected V-shaped cleft in the bilateral feet of the fetus and a history of similar deformity in the first child [8]. Lévy et al. reported a triplet gestation in which one fetus showed ectrodactyly with radial agenesis at 10-week ultrasonography, following which selective fetal reduction was performed [9]. Chauhan et al. described antenatal detection of severe ectrodactyly with a single digit in each limb, with a positive maternal history of similar deformities [5]. Mandal et al. reported unilateral SHFM in an 8-year-old boy involving the right hand, characterized by a small palm, rudimentary digits, and absence of bony structures distal to the carpal bones [2].

Ashi et al. described SHFM in siblings presenting with bilateral hand and foot deformities, normal growth and development, and no prior family history of congenital anomalies [10]. A retrospective review of ultrasound records from fetal care centers in the USA and China (2002–2012) identified 10 pregnancies with sonographic features suggestive of SHFM between 15 and 29 weeks’ gestation. Seven pregnancies were electively terminated because of severe malformations, while three continued to term delivery [11]. Parents at risk should be offered genetic counselling and, when appropriate, pre-implantation or prenatal genetic testing. They should also be counselled about the possibility of the same in future siblings.

There is no effective non-surgical therapy for ectrodactyly. Prosthetic devices may provide cosmetic benefit; however, surgical reconstruction remains the mainstay of management and should be individualized, prioritizing functional and

cosmetic improvement. Management should be multi-disciplinary, involving various surgical specialties like Orthopedics and Plastic Surgery. Briefly, the surgery involves cleft closure, syndactyly release, osteotomies for alignment, and soft tissue balancing [12]. Rehabilitation with appropriate physiotherapy is equally important for improving functional outcomes. The surgical team should counsel the family regarding available options, optimal timing, and the sequence of procedures.

CONCLUSION

SHFM represents a diverse group of limb malformations that may affect one or multiple extremities and can occur alone or within a syndrome. Early diagnosis allows families to make informed decisions, including the option of pregnancy termination in severe cases. Identifying the specific genetic mutation in affected relatives enables accurate counselling, targeted testing, and consideration of pre-implantation genetic diagnosis for future pregnancies.

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Funding: None; Conflicts of Interest: None Stated.

How to cite this article: Gautam G, Choudhary M, Priyadarshi M, Chaurasia S, Singh P, Basu S. Isolated Ectrodactyly in a Late Preterm Neonate: A Case Report. Indian J Child Health. 2026; 13(4):53-55.