

Familial Ectrodactyly: a Rare Report Of Lobster-Claw in a Malay Family

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Abstract

We describe a case of ectrodactyly in isolated non-syndromic form claw in three living generation of a Malay family which presented as autosomal dominant pattern of inheritance with sporadic occurrence. To our knowledge, this is the first report of such case in Malaysia.

A three-year-old boy with claw toes presented to Paediatric Orthopaedic Unit accompanied by his mother and grandfather. He is an active child with normal intellectual. Neither facial dysmorphism nor cleft lip and palate was observed. His right tibia is shorter than the left one and his ankle is totally dislocated posteriorly. His right foot has only one ray and his right ankle joint is in 100° equinus. For his left lower limb, the leg and ankle are normal. However, the left foot is bifid with two lateral and one medial ray. He can walk on the stump of distal right ankle but with short limb gait. He prefers to walk in kneeling position as he finds it a bit faster and that he can move freely. Other systemic reviews are unremarkable.

His mother was born with the similar deformity on the feet and ankle. She underwent below knee amputation at the age of 6 and has been using prosthesis since after corrective surgery failed. The grandfather has central splits in both hands and bifid femur bilaterally. Only one ray present on each foot.

The prognosis for most people with ectrodactyly syndrome is very good. Various cosmetic prosthesis, amputation or reconstructive surgery might help, depending on the type of deformity and as per case basis. Multidisciplinary care would contribute significantly to improve the survival and the quality of life of a child with congenital musculoskeletal malformations such as ectrodactyly.

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Introduction

The word ectrodactyly is derived from Greek words; 'ektroma' which means abortion and 'daktylos' which means finger.¹ It is characterised by the absence of one or more fingers or toes with deep median clefts. Also known as split hand/foot malformation (SHFM), this congenital is often described as lobster claw due to its appearance.²

Ectrodactyly may be associated with syndactyly, aplasia or hypoplasia of phalanges, metacarpals and metatarsals.³ The incidence is

rare, generally is reported in 1 per 90,000 live births.⁴ It may present either as an isolated case or associated with a syndrome. In syndromic form, ectrodactyly occurs in combination with other anomalies, such as ectodermal dysplasia, cleft lip and palate, genitourinary and other anomalies of ectodermal origin.^{5,6}

Often in genetic inheritance, ectrodactyly cases present as autosomal dominant pattern with reduced penetration.⁷ Infrequently, it exhibits autosomal recessive^{3,8} and X-linked forms of transmission.⁹ It may also be caused by chromosomal deletions and duplications.¹⁰

We describe a case of ectrodactyly in isolated non-syndromic form claw in three living generation of a Malay family which presented as autosomal dominant pattern of inheritance with sporadic occurrence. To our knowledge, this is the first report of such a case in Malaysia.

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Case Report

A three-year-old boy with claw toes presented to Paediatric Orthopaedic Unit. He was accompanied by his mother who was wearing patella-tendon-bearing prosthesis and his grandfather who also has lobster claw deformity on all four limbs. (Figure 1, 2, 3, 4, 5, 6).



Figure 1. the three generations with split hands and feet malformation.

He was born at term via spontaneous vertex delivery after an uncomplicated pregnancy to a 28-year-old mother and a 32-year-old father. He is the product of non-consanguineous marriage. He is the only living child as his younger brother had intrauterine death at eight months and was born with the same deformity over bilateral lower limb.

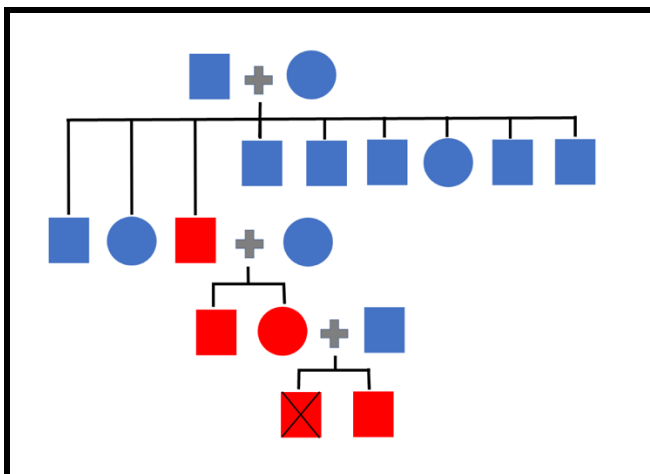


Figure 2. The family tree that shows autosomal dominant pattern of inheritance with sporadic occurrence of ectrodactyly.

On examination, he is an active child with intellectual appropriate to his age. No facial dysmorphism was observed. His facial bone

grossly looks normal with the absence of cleft of lip palate that is usually observed in ectrodactyly, ectodermal dysplasia with cleft palate (EEC) Syndrome.² The child mobilises by walking on both knees. Callosities were observed over anterior aspect of his knees. His right tibia is shorter than the left one and his ankle is totally dislocated posteriorly. Regardless, he still can weight bear on distal end right tibia fibula. His right foot only has one ray and his right ankle joint is in 100° equinus. For his left lower limb, the leg and ankle are normal. However, the left foot is bifid with two lateral and one medial rays. Otherwise, the heel is well formed and the child is able to fully weight bear on the left foot. He can walk on the stump of distal end tibia and fibula on right side but with short limb gait. He prefers to walk in kneeling position as he finds it a bit faster and that he can move freely. Both of his knees are supple, stable and can flex fully. Other systemic reviews are unremarkable.



Figure 3. The child is able to weight bear on distal end right tibia fibula. With kneeling, the limbs become symmetrical, and the child becomes more mobile. Evidence of callousity over bilateral anterior knee was observed due to habitual kneeling walking.

Concurrently, the child is also having possible familial hearing impairment from its paternal side. His grandmother, father, three uncles and two cousins are also affected.

His mother was born via emergency

caesarean section operation for pre-eclampsia and breech presentation. The mother had an elder brother who was stillborn. He was born with claw hands and feet at term. The mother is now using bilateral transtibial prosthesis. She was born with the similar deformity on the feet and ankles but has normal hands and upper limbs. An attempt to correct the position of ankle was done when she was four using Taylor Spatial Frame external fixator but failed. Her parents then decided that below knee amputation was the best option for her to fit in the prosthesis. She started using prosthesis at the age 6. Her education level is until college.



A



B

Figure 4. (A and B) Only one ray presents on right foot and the ankle is distorted with posterior dislocation ankle and equinus deformity ipsilateral side. The left foot shows typical split foot with the absence of median rays.

The grandfather was born via natural method at home. Out of nine siblings, he is the only one with deformity over bilateral upper and lower limbs. There was no history of consanguinity or any other congenital malformation as he denied the existence of similar deformity in elder generations. His parents were rubber tappers and there is no history of exposure to radiation in both parents. He came from low-income family and had completed primary school education. He earned a living by selling newspapers and currently is independent in doing daily living activities. He is

mobilising with wheelchair and utilize a specialised three-wheel motorbike to move from one place to another. He got married to his wife at the age of 30 and is blessed with one living child. The elder son was born stillbirth and had the same deformity as he on all four limbs.



Figure 5. The mother is using bilateral patella-tendon-bearing prosthesis.



A



B

Figure 6. (A and B) The grandfather with ectrodactyly of bilateral hands and feet. He also has associate bifid femur bilaterally.

During examination, he sat on his wheelchair and was able to wheel on his own. He has central splits in both hands. On the left hand, there was absence of middle finger while the thumb and the index fingers were fused together.

Otherwise, the little and the ring fingers were normal. On the left hand, the middle and the ring fingers were absent. The other fingers appeared normal. For the lower limbs, he has bifid femur bilaterally with one of the femur articulating with the knee joint however the knee joint was stiff in hyperflexion position. The bilateral ankle joint was in 100° equinus and dislocated posteriorly. Only one ray present on each foot.

Discussion

This case was presented to us as the parents requested for option of treatment to improve mobilisation of this child. Their wish is for the child to walk as a normal person, go to school, and ultimately, earn a living. Being affected themselves, the parents understand the needs to get the best treatment as soon as possible.

Early physical and occupational therapy can help those with ectrodactyly to adapt to learn and become fully functional even with the deformities that they have. In this child, although the right leg is shorter than contralateral side, he learnt to use the knees to walk as the length of femur is the same, so the knee is at level. This allows him to walk faster and be as active as other normal children.

One of the issues with ectrodactyly is social stigmata. Due to physical abnormality over the hands and feet, it is difficult to hide them. The psychology of a child might also be affected, which may have negative impact on their mental development.

As the left foot is stable, it is best not to do any procedure to it now. Custom made shoe can be used to give better external appearance. While for the right foot, options of treatment for his lower limb are custom prosthesis fitting or surgery; which can be divided into two; reconstructive or destructive surgery.

In regard to custom prosthesis for the child, the design will be bulky and externally. Although the appearance will not be pleasing, the child will not be subjected to any surgery. For option of reconstructive surgery, an attempt can be made to correct the dislocated and equinus over right ankle and followed by arthrodesis of the right ankle. The shortening of right leg can be addressed by distraction and lengthening of affected tibia. However, the downside is the long procedure duration and the possibility of surgery

failure. Given that the mother had gone through this procedure before, she is not keen for her son to go through similar experience only to fail.

For option of destructive surgery, as unnerving as it may sound, it still has its advantages. The rehabilitation programme could be accelerated once the wound recovers, but the outcome of the surgery is permanent. Early rehabilitation could help the child to adapt early and learn to function close to normal. With prosthesis, the deformity could be hidden grossly, for instance, by using long pants. This will help in reducing mental trauma to the child. In this case, since the mother had undergone bilateral transtibial amputation which had improved her lifestyle, she is hoping the same for her son.

Conclusions

The prognosis for most people with ectrodactyly syndrome is very good. Various cosmetic prosthesis, amputation or reconstructive surgery might help, depending on the type of deformity and as per case basis. Multidisciplinary care involving Paediatrician, Orthopaedic Surgeon, Rehab Physician, Physiotherapist and Occupational Therapist would contribute significantly to improve the survival and the quality of life of a child with congenital musculoskeletal malformations such as ectrodactyly.

Declaration of Interest

The authors report no conflict of interest.

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