Picture stories

A case of hand-foot-genital syndrome with sporadic inheritance: first reported case from Sri Lanka

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Introduction
Hand-Foot-Genital Syndrome (HFGS) affects limbs and genitourinary system where most cases are familial with autosomal dominant inheritance[1]. We report a boy with HFGS with sporadic inheritance.

Case report
A 4 year and 10-month-old boy presented with difficulty in wearing slippers due to deformity in the big toes. He was comfortable wearing shoes. He also had short thumbs. There was no family history of similar deformities. He was born to non-consanguineous parents by uncomplicated vaginal delivery. The deformity of the toes was noted at birth. There was no family history of similar deformities. He achieved his developmental milestones at an appropriate age. He did not have significant medical illnesses in the past such as urinary tract infections.

His height was 102 cm (10th centile) and weight was 14kg (3rd centile). There were bilateral short first metatarsals with proximally placed big toes (Figure 1) and bilateral short first metacarpals with proximally placed thumbs, short poorly developed thenar eminences and bilateral clinodactyly (Figures 2 and 3). There were reduced palmar creases in the proximal palm. Abdominal examination did not reveal organomegaly. Penis showed chordee. Cardiovascular, respiratory and neurological examinations were normal.

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Examination of both parents did not reveal evidence of HFGS.

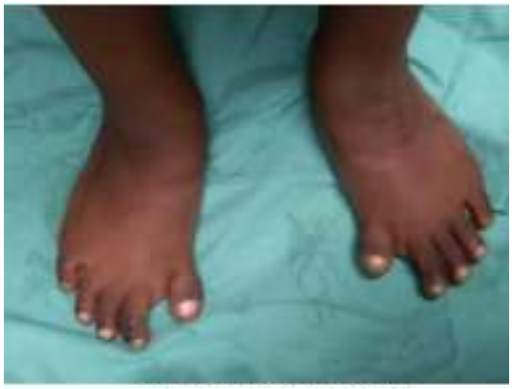

Figure 1: ***Bilateral feet***

Figure 2: Bilateral hands with short thumbs and clinodactyly

Figure 3: Child's hands compared with mother's hands. Clinodactyly, short proximally placed thumb and small thenar eminence noted in the child

Bilateral hand radiographs showed short first metacarpal with clinodactyly (Figure 4). Bone age was 14 months to 30 months which was significantly delayed compared to chronological age of 58 months. Radiographs of bilateral feet showed short

first metatarsal, small triangular first proximal phalanx and medial deviation of metatarsals (Figure 5).

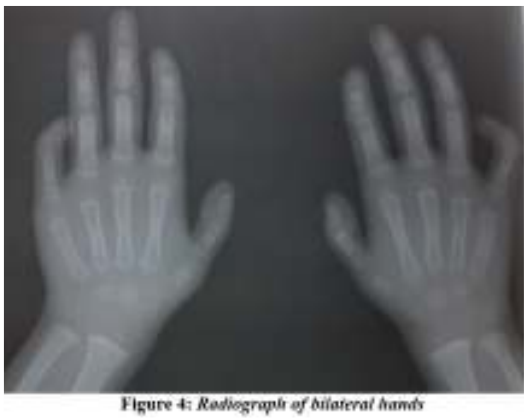

Figure 4: *Radiograph of bilateral hands*

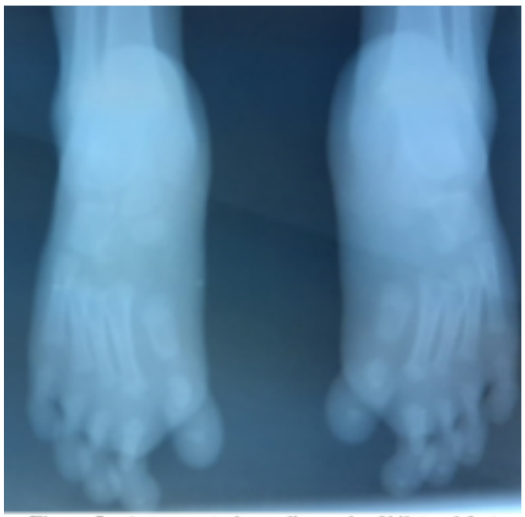

Figure 5: *Anteroposterior radiograph of bilateral feet*

Serum free thyroxine level was 1.09 ng/dL (normal range 0.9- 1.72 ng/dL) and thyroid stimulating hormone level was 1.39mU/L (normal range 0.8 – 6.23). Ultrasound scan of the genitourinary system did not show any abnormalities. Echocardiography showed a structurally normal heart. Paediatric surgical follow up was arranged for chordee of penis.

Discussion

HFGS is a rare autosomal dominant disorder affecting limbs and genitalia[1,2]. There were less than 25 familial cases and five sporadic cases reported as at 2017[2]. A thorough literature search did not reveal any reported cases from Sri Lanka. This is the first reported case of HFGS in Sri Lanka. Our patient had short first metacarpal and metatarsal, clinodactyly and chordee of the penis which are described in HFGS[3]. Delayed bone age is a known feature of HFGS[3]. Mutations in *HOXA13* gene in short arm of chromosome 7 is responsible for HFGS[3]. Genetic testing could not be carried out due to financial and logistical constrains. Hand or foot surgery is not usually needed[1]. It is important to look for features of a syndrome when several dysmorphic features are present. It will help to screen the patient for possible complications associated with the syndrome.

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