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MANAGEMENT OF POST-AXIAL POLYDACTYLY WITH DUPLICATION IN A CASE OF BARDET BIEDL SYNDROME

Plastic Surgery

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ABSTRACT

Bardet Biedl Syndrome is an autosomal recessive disorder characterized by multisystem involvement. Typical clinical manifestation includes Central Obesity, Polydactyly, Poor IQ, Hypogonadism, Renal and retinal problems. We present a 16-year-old girl who was brought by her mother for management of extra digits in both hands and feet. Syndromic association was established by features of Hypogonadism, Diabetes Mellitus, Hypothyroidism, Obesity and Learning Disability. Patient was evaluated to rule out other congenital anomalies and medical treatment started for correction of endocrinological disorders. Correction of Bilateral Ulnar polydactyly and duplication of little finger in right hand performed after control of diabetes. Post-operatively patient hand functions improved with physiotherapy.

KEYWORDS

Bardet Biedl Syndrome; Hypogonadism; endocrinological disorders; Duplication; Post-Axial Polydactyly

INTRODUCTION

Bardet biedl syndrome is a rare congenital anomaly which requires a thorough evaluation of the systemic involvement characteristic to the syndrome. Patients usually seek for management of hypogonadism, polydactyly. [1,2] Problems associated with ulnar polydactyly include both functional and cosmetic appearance. Hence early correction of polydactyly is essential for improvement of quality of life.

Case Report: A 16-year-old girl, right handed individual was brought to the OPD by her mother seeking correction of polydactyly and irregular cycles. Patient is a known case of hypothyroidism on medical management for 2 years. Patient was initially admitted under endocrinology for management of hypogonadism and was also diagnosed to have Diabetes Mellitus. Bardet biedl syndrome was established on the basis of following clinical features:

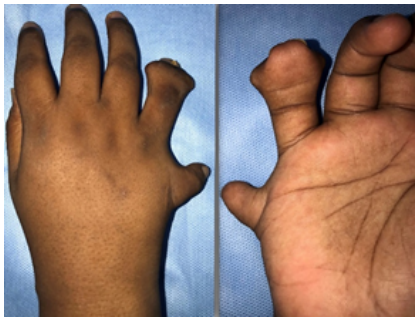
1) Obesity
2) Post-axial polydactyly
3) Hypogonadism
4) Low IQ & learning disability
5) Diabetes Mellitus

Following control of Blood sugars patient was referred to the Department of Hand Surgery for correction of the hand anomaly. The patient faced problems due to the polydactyly especially while resting the hand on table while writing exams. She was subjected to criticism at school due to the appearance of her hand. The difficulty in wearing ornaments such as bangles was yet another concern of the mother.

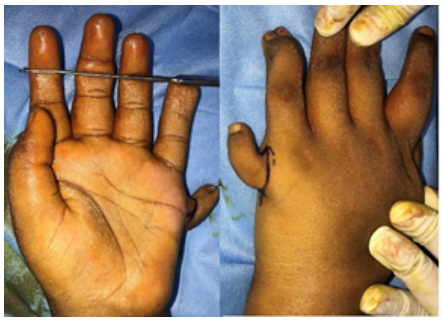
Examination:

The patient was obese with a BMI of 31.3. She had poor visual acuity and Low IQ.

Examination of her hand revealed B/L Type A Ulnar Polydactyly with duplication of little finger and synonychia. The little finger of the right hand also had a deviational deformity. The palmar surface revealed well-developed creases at each joint. The extra digit on the right hand had a bony origin from the 5th metacarpal and there was weak flexion of the digit at IP joint (Fig 1).



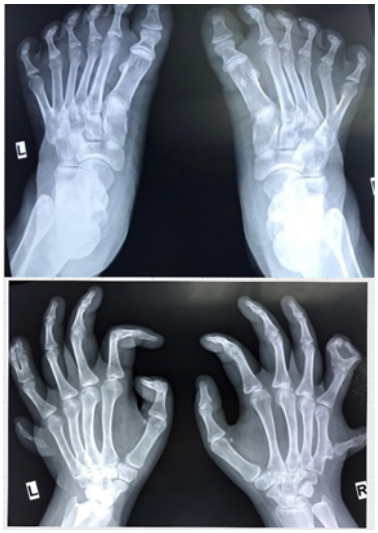
The extra digit on the left hand was freely mobile and had no active flexion or extension at the IP joints (Fig 2). There was sensation in the excess digits with a normal capillary refill time. The patient also had polydactyly in her feet.



INVESTIGATIONS

X-Ray of both hands revealed Ulnar polydactyly with duplication in the little finger. The polydactylous digit had a bony origin from the shaft of the 5th metacarpal of the right hand. The little finger had a bifid middle phalanx with a deviation of the terminal phalanx and complex syndactyly. The polydactyly in the left hand had no bony attachments with the metacarpal. The little finger had a bifid Middle phalanx with no deviation and simple syndactyly.

X-Ray of the foot showed a bifid 5th metatarsal and a well-developed 6th toe without any deformity (Fig 3).



DIFFERENTIAL DIAGNOSIS

- **Ellis-van Creveld syndrome**: Differentiated from BBS by the absence of retinitis pigmentosa, obesity, and hypogonadism.
- **Meckel-Gruber syndrome:** Distinguished from BBS by the absence of RP and the presence of encephalocele.
- **Alström syndrome:** It is characterized by relative preservation of cognitive function and the absence of polydactyly
- **McKusick Kauffman Syndrome:** Triad of hydrometrocolpos (HMC), postaxial polydactyly (PAP), and congenital heart disease (CHD). Requires genetic testing to differentiate.
- **Joubert Syndrome:** Brain abnormalities with "molar tooth sign" in MRI are hallmark features

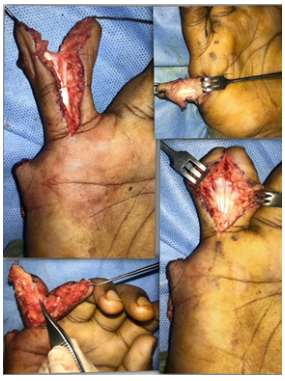
TREATMENT

The patient is a known case of hypothyroidism on treatment with 75mcg of thyroxine for 2 years. The patient underwent evaluation for hypogonadism and was treated for the same with hormonal supplements progestin and oestrogen. The patient was recently diagnosed to have diabetes and was started on insulin for pre-operative control of hyperglycaemia.

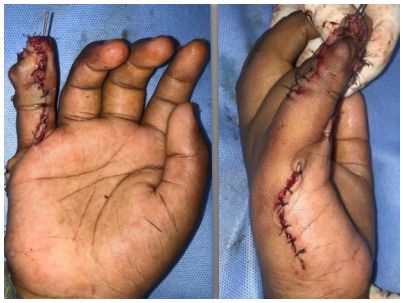
The patient was planned for surgical correction of polydactyly in both the hands and correction of duplication in the right hand because of the deformity.

The patient was taken up under general anaesthesia in a supine position and tourniquet control. The right hand was operated first. A circumferential incision was made around the polydactyly and extended to the ulnar border of the hand. The neurovascular bundle and accessory tendons were retracted and divided after ligating the vessels. The ligamentous attachments with the bony origin on the 5th metacarpal were released and the digit amputated. The bony spur was chiselled to achieve a smooth contour to the 5th metacarpal and provide a uniform surface for the hand to rest upon.

Next, the syndactyly in the little finger was released after raising skin flaps. A wedge osteotomy of the bifid middle phalanx was done. The radial side Middle phalanx was retained and ulnar side Middle and terminal phalanx along with nail complex were sacrificed after retaining the skin flaps and ulnar collateral ligament along with its bony attachment to the base of the middle phalanx. Wire encirclage was done with a 24-gauge Stainless steel wire to fix the portion of the middle phalanx and ulnar collateral ligament. The accessory FDP tendon was sutured to the radial side FDP tendon (Fig 4).



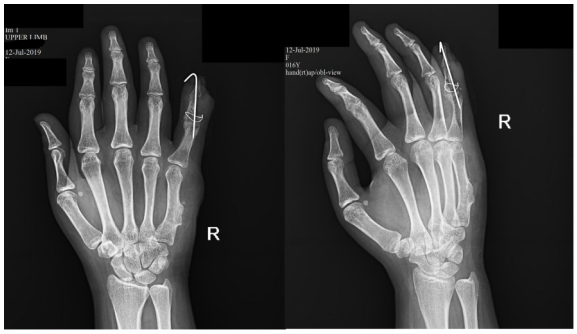
The deviation was corrected passively and fixed internally with a single 1.25mm k wire. The raw area on the ulnar side was covered with the filet flap retained from the duplication (Fig 5).

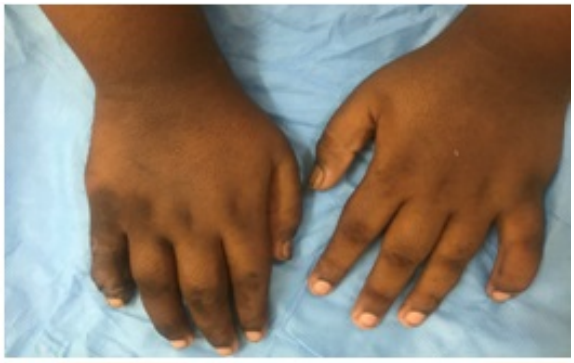


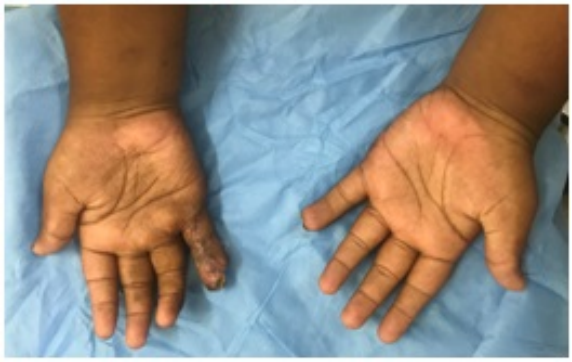
The polydactyly in the left hand was excised after ligating and dividing the neurovascular bundle. There were no bony attachments or accessory tendons to the digit. The patient was planned for correction of duplication of the little finger in the left hand at a later date. Post-operatively the patient was monitored for the viability of the fillet flap and digit in the right hand. Pain tolerance of the patient was poor and hence required analgesics for a longer duration. The patient and her mother were educated about maintaining local hygiene and control of diabetes. The patient was also advised for review periodically for her medical issues.

OUTCOME AND FOLLOW-UP

The patient was followed up for 6 months. Post-operatively there was tip necrosis of the fillet flap used for the ring finger of the right hand. With regular dressing, the wound healed well. Sutures were removed on Post-operative day 10 and K-wire removed on Post-operative day 21 (Fig 6,7).







The initial stiffness of the Proximal interphalangeal joint of right little finger improved with Physiotherapy. The patient was referred to occupational therapy to improve her hand function. The patient was also closely monitored for glycaemic control and thyroid function.

DISCUSSION

Bardet-Biedl syndrome is a rare ciliopathic pleiotropic autosomal-recessive disorder named after Georges Bardet and Arthur Biedl observed a complete form of this syndrome for the first time. [3,7] There is a slight male predominance with consanguinity is a risk factor. [3,7]

The incidence in Kuwait and Newfoundland has been higher due to consanguinity. [4] This syndrome has been mapped to 20 genes (BBS1-21) which code for proteins involved in ciliary biogenesis and functions. [4] Mutation in these genes affects the structure and function of cilia. The most common defective gene is the BBS1 gene located on the long arm chromosome 11q132,5,6.

Early diagnosis is made based on clinical examination and establishment of clinical criteria. [2]

Patients who had 4 primary characteristics or 3 primaries and 2 secondary criteria were identified as BBS [Table 1]

Table 1: Clinical criteria

Primary Features	Secondary Features
Retinal dystrophy	Speech delay/disorder
Post–axial polydactyly	Developmental delay
Truncal obesity	Craniofacial dysmorphism
Hypogenitalism	CVS anomalies
Renal abnormalities	Hepatic involvement
Learning disabilities	Anosmia, ataxia, mild hypertonia

BBS has an early onset of blindness, obesity, hypertension, and diabetes mellitus. Renal impairment is the major cause of morbidity/ mortality in these patients. Management is specific to the symptoms and requires long-term surveillance and coordinated effort of paediatrician, plastic surgeon, endocrinologist, dentist, speech pathologist, ophthalmologist, cardiology, and nephrologist.

Diabetes Mellitus is a common feature observed in these patients which requires a regular follow up. Genetic counselling is advisable to the family members. Baseline investigations include Renal and liver function test, ECG, and Ultrasound abdomen to check for renal anomalies. Electroretinogram, IVP, and ECHO are also helpful. [6] Assessment of vision and speech are essential to educating the patient. Molecular Genetic diagnosis and multigene panel often helps in disease confirmation. [4,5]

Our patient satisfied the above-mentioned criteria and was initially under medical management for uncontrolled diabetes, hypothyroidism, and hypogonadism. The patient was referred for the management of polydactyly with duplication in both hands since it hindered activities such as resting the hand on the table while writing. The patient underwent removal of Type A Postaxial polydactyly in both hands with simultaneous correction of duplication in the little finger of the right hand. Post-operatively patient was followed up for 6 months. The patient and her parents were satisfied with the outcome.

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