

Oral-Facio-Digital Syndrome in a Nigerian Infant : a Case Report

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Abstract

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Baby YS, a 72-hour-old term male baby, was admitted on account of an abnormally small tongue, multiple digits and fast breathing since birth. Fast breathing started shortly after home delivery which was conducted by a nurse. The mother had drained liquor for three days prior to delivery. Examination revealed an active baby, crying fairly audibly, with rudimentary, multi-lobulated tongue, irregular gum, high-arched palate, fleshy outgrowth in both nostrils, and low set ears. There were also multiple digits on all the limbs, six each on the upper, and seven on the lower limbs with syndactyly of the two right big toes; there was a single palmar crease bilaterally. Anthropometry measurements were all within normal limits for a term baby. His respiratory rate was 80 cycles per minute, with inspiratory stridor, intercostal recessions and pectus carinatum. Auscultation revealed transmitted sounds. In the urogenital system, there was micropenis, with a stretched penile length of 1.2cm. Abdominal and central nervous systems were normal for the age. Initial assessment was that of multiple congenital anomalies and respiratory distress with risk for sepsis. Investigations requested immediately included X-ray of the limbs, chest X-ray, and full blood count. Antibiotics, intravenous fluids, and intranasal oxygen were commenced. His respiratory distress gradually improved over the next three days, and he began to tolerate expressed breast milk via cup and spoon. He was well enough to be discharged after six days of admission. However, he defaulted from follow up and died at home at the age of three months. Extensive literature search revealed a rarity of the malformations.

Introduction

CONGENITAL malformations are structural abnormalities of any type present at birth.¹⁻⁶ They could be macroscopic, seen on the surface or be within the body, and may be major or minor.^{1,2} Major malformations are defined as those with serious medical, surgical and cosmetic consequences.³ Global incidence ranges between 2 and 5 percent^{2,3} with wide variations from place to place. As birth trauma, infections and other causes of perinatal deaths came under control in many industrially developed countries, congenital malformations have come to contribute substantially to perinatal morbidity and

mortality.⁴ Spontaneous abortions in early gestation are often due to genetic disorders associated with complex congenital anomalies that are incompatible with life.⁵ A generally accepted principle is that the presence of three or fewer minor malformations is usually not significant, however, the presence of more than three minor malformations should alert the observer to an increased risk of a major malformation.^{1,6}

The aetiology of congenital malformations is unknown in 50-60 percent of cases; they are of multifactorial origin in 20-25 percent of cases, while environmental factors, chromosomal aberrations and gene mutations are collectively responsible for the remaining 20-25 percent.¹ Smith, who is regarded as the father of dysmorphology,² highlighted four types of structural defects that can result in a chain of defects at birth as malformation, deformation, disruption and dysplasia.^{1,6} A number of congenital malformations are already well established,¹⁻⁶ however, new anomalies may be seen with features dissimilar to earlier ones.³ In evaluating a child with

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dysmorphic features, Smith^{2,6} suggested the following four steps (1) gathering information with detailed history, (2) interpreting the anomalies from a viewpoint of developmental anatomy, (3) attempting to arrive at a diagnosis when feasible, and (4) giving prognosis, outlining management and counseling accordingly. Regarding nomenclature, he recommended that in a single malformation, a descriptive adjective be used with the name of the structure;⁶ however, in naming an entity with multiple malformations, the use of eponyms is allowed though not in the possessive form.

Our intent is to report an unusual combination of multiple malformations, and to call for a more concerted national effort in organizing and subsidizing the cost of evaluation and management of children with dysmorphic features.

Case report

Baby YS, a 72-hour-old term male baby presented to the Emergency Paediatric Unit (EPU) of the Federal Medical Centre, Bida on account of abnormal tongue, multiple fingers and toes and fast breathing from birth. The mother had no formal antenatal care and delivery was said to have been supervised by a nurse at home. There was drainage of liquor for three days prior to the onset of labour. The baby was said to have cried spontaneously after delivery. He was commenced on breast feeding shortly after delivery but was unable to suckle well. There was no history of force feeding or choking while being fed. No history of tobacco smoking, ingestion of alcohol, exposure to radiation or unprescribed drugs antenatally by the mother, however ingestion of traditional medicine predated and transcended pregnancy and delivery. No history of fever or rash during pregnancy. The family setting was monogamous; the mother had a first trimester spontaneous abortion in the preceding pregnancy.

The baby was the second child in the family, the older sibling being a five-year-old normal and healthy boy. There was no similar condition in the families of both parents. The father was a security officer at Kuta, in Shiroro Local Government headquarters in Niger state, while the mother was a full-time housewife. The father, aged 42 years, had secondary education certificate, while the mother aged 29 years, had no formal education.

Examination revealed a conscious child, pink in room air, afebrile (temperature 36.9°C), with subconjunctival haemorrhage in the left eye, bilateral periorbital hyperemia, low-set ears and micrognathia (Fig. 1). Anthropometry was essentially normal with occipito-frontal circumference at 33.0cm, length at 47.5cm, arm span 47.4cm, upper segment 24.5cm,

lower segment 23.5cm and weight of 3200g. More detailed findings included the following:

Mouth: (Figs 2 and 3)

- Marked hypoglossia
- Rudimentary tongue with micro lobulations
- Irregular, high arched palate

Nose

- Fleshy outgrowth within both nostrils (Figs 2 and 3)

Hand: (Figs 4a and 4b)

- Single palmar crease bilaterally
- Polydactyly

Feet (Fig 5)

- Pre and post axial polydactyly with complete duplication of the right hallux, and polysyndactyly of the left with partial duplication of the hallux.

Urogenital system: (Fig. 6)

- Micropenis (stretched length of 1.2cm)
- Normally descended testes
- Scrotal sac with good rugae

Respiratory system

- Tachypnoea with inspiratory stridor
- Respiratory rate of 80 cycles/minute
- Intercostal recessions with pectus carinatum
- Transmitted sounds on auscultation, no crepitations

Cardiovascular system

- Pulse was normal in volume and rhythm
- Heart rate of 160 beats/minute
- Apex beat located in the 4th left intercostal space, mid-clavicular line
- Normal heart sounds with no murmurs

Gastrointestinal system

- Abdomen was full and moved with respiration
- Umbilical stump was well secured
- Liver was tipped, liver span was 6cm, spleen was not palpable and the kidneys were not ballotable
- Bowel sounds were present and normoactive

Central nervous system

- Conscious, active, and well flexed
- Appropriate response to stimulation
- Primitive reflexes were present and normal

An assessment was made of a neonate with multiple congenital anomalies who was at risk for sepsis on account of maternal prolonged rupture of membrane.

Investigations requested included X-ray of the limbs which was not done on account of financial constraints; chest x-ray which showed normal cardiac

silhouette with few patchy lung opacities, urinalysis that was normal, and urine culture which grew no pathogen. Abdominal ultrasound showed a normal scan and the full blood count was essentially normal for age with a packed cell volume of 48 percent, white cell count of $5.2 \times 10^9/L$, with 60 percent neutrophils and 40 percent lymphocytes.

Treatment offered included intravenous (iv) 4.3 percent dextrose in 0.18 saline made up to 10 percent dextrose administered at 90mls/kg, iv cefuroxime and intramuscular (im) gentamicin. Other supportive treatment including suctioning as required, was also carried out.

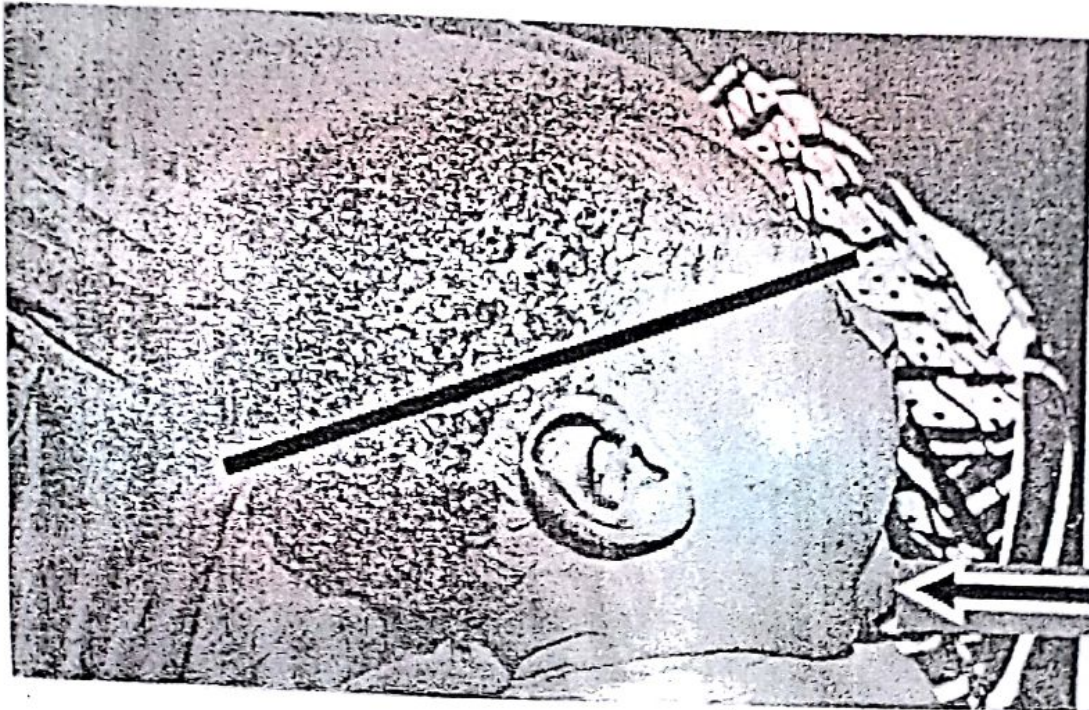


Fig. 1. Low set ear and micrognathia (arrow)



Fig. 2. Marked microglossia with microlobulations of the rudimentary tongue

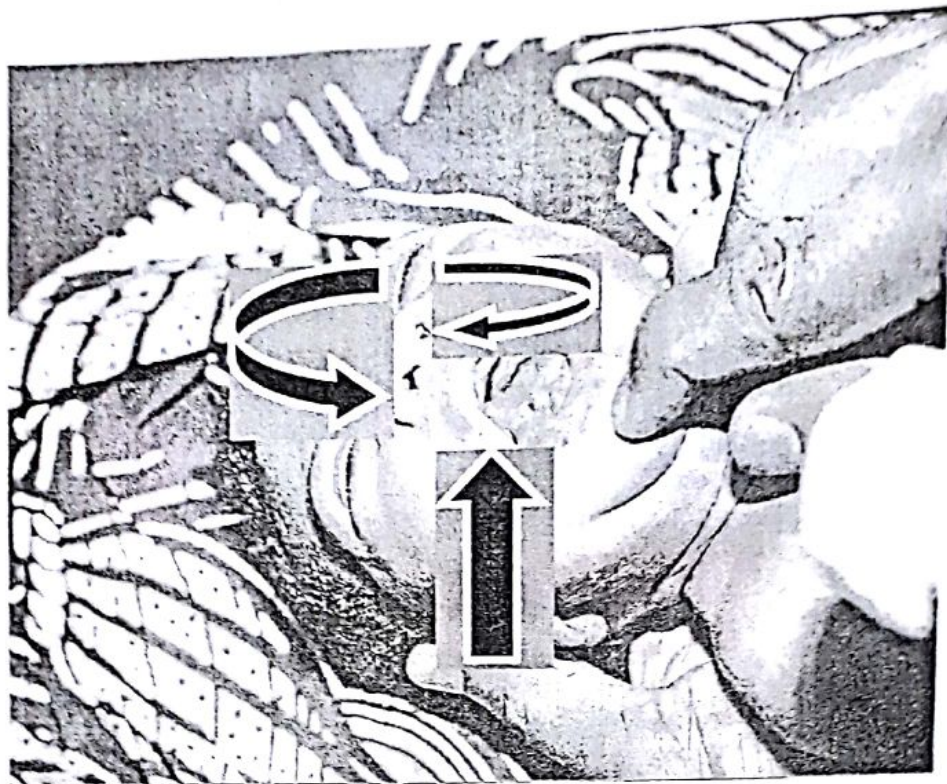


Fig. 3: Marked microglossia with fleshy outgrowth in the nostrils



Fig. 4a: Single palmar crease with bilateral polydactyly

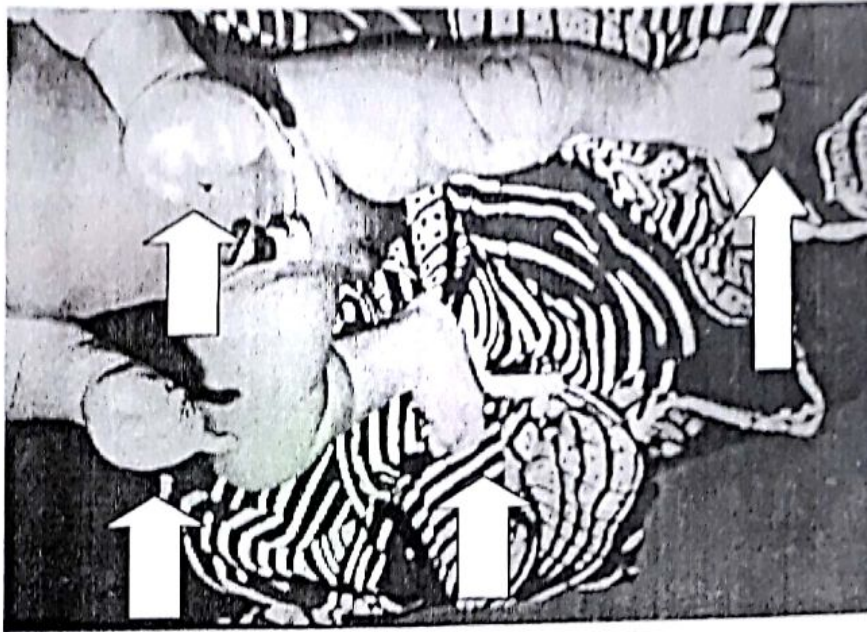


Fig. 4b: Polydactyly on all limbs

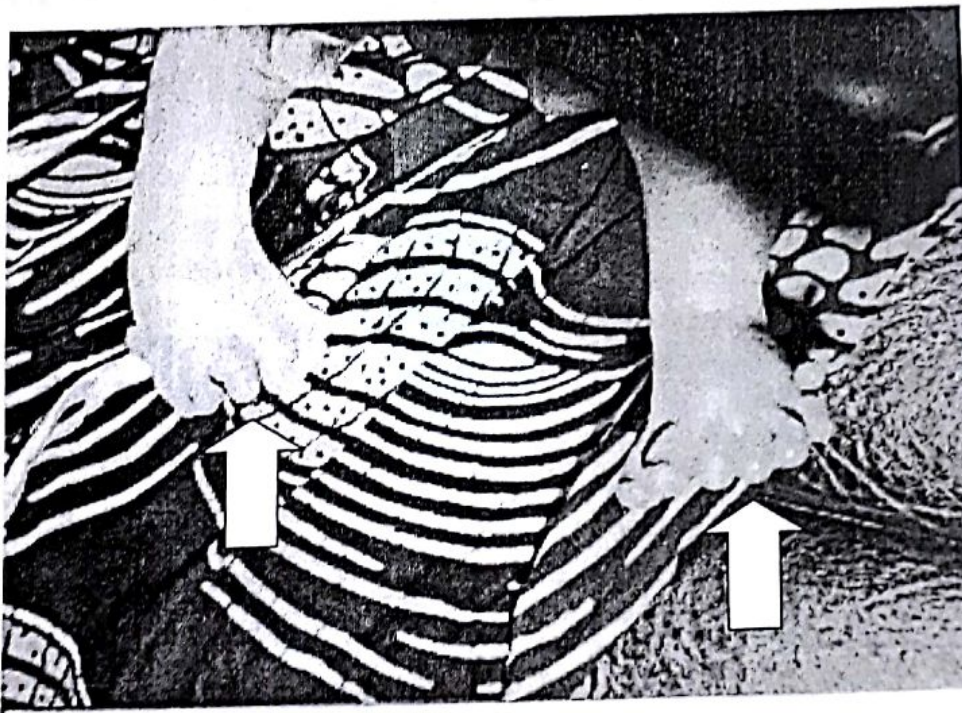


Fig. 5: Pre and post axial polydactyly right hallux, and polysyndactyly on the left.

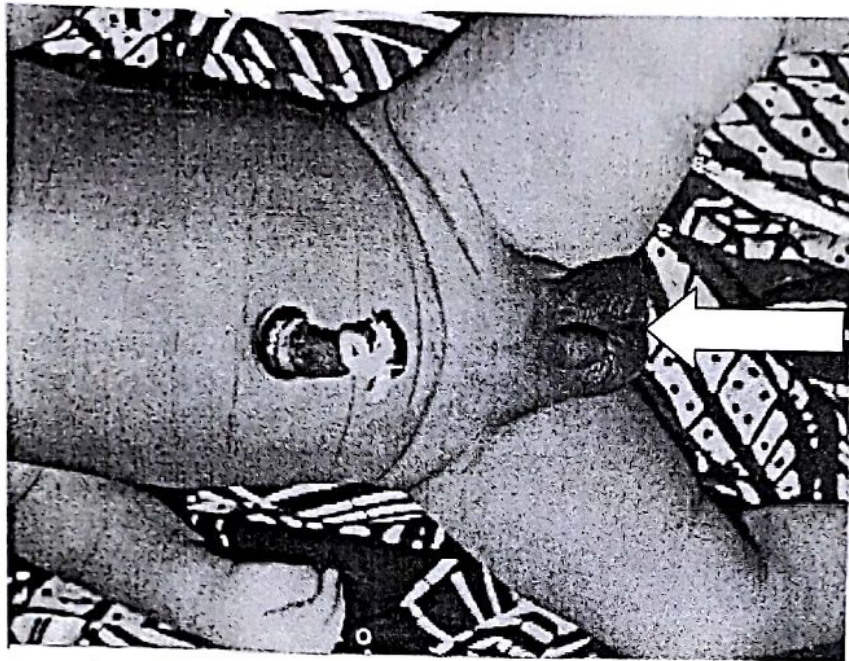


Fig. 6: Micropenis

The respiratory distress progressively resolved. Cup and spoon feeding with 20mls of expressed breast milk (EBM) two hourly (75 mls per kilogram per day) was commenced on the 4th day of admission, while the volume of iv fluid was reduced to 40mls per kilogram per day, and was finally discontinued on the fifth day when the feed was increased to 24mls, two hourly (90 mls per kilogram per day) with the baby tolerating the feeds well.

At this stage, the father started agitating for discharge and was counseled. However, he then expressed his concern about the baby's tongue and asked if another tongue could be provided. The baby was eventually discharged after six days on admission, to be seen at the follow-up clinic after one week. However, the parents did not bring the child for the follow-up visit. Three weeks later, our social work unit was successful in tracing the family without seeing the baby, but was told that child had been taken to an undisclosed place and got an assurance that the baby would be brought for follow up, two weeks later. As this did not happen, a repeat tracing effort was carried out after another two months and this revealed that the baby had died at the age of three months, a week before the second visitation.

Discussion

Congenital anomalies of the tongue could be as minor as ankyloglossia, fissured tongue, scrotal tongue⁷ or aberrant frenulum,² or could be a major one causing severe discomfort to the child and

affecting his/her normal living pattern.^{6,7} Hypoglossia is one of the rare congenital anomalies manifested by the presence of a small or rudimentary tongue.^{7,8} Being an organ instrumental to effective suckling in the newborn, a major abnormality of the tongue is likely to be associated with significant feeding challenges as seen in our case. A review of the literature shows that hypoglossia though rare, with a prevalence of 1 per 250,000, is often associated with other cranio-facial anomalies such as microstomia, micognathia, partial or complete anodontia, cleft palate, facial asymmetry and cranial nerve palsies.^{7,9} Earlier workers have suggested that one out of every 100 patients with cleft palate had one form of orofaciogigital syndromes (OFD) or the other. Limb defects are also commonly present in such babies with anomalies ranging from absence of distal phalanx, partial absence of the limb with or without syndactyly, to total adactyly.^{7,10,11} All the four limbs may be affected.¹⁰⁻¹² In consonance with earlier reports,^{7,9-13} our case had involvement of all limbs, micrognathia, syndactyly with polydactyly on all the limbs with full duplication of the left big toe and partial duplication of the right big toe. Our case is considered a variant/form of OFD which consists of a group of heterogeneous disorders characterized by abnormalities in the oral cavity, face and digits. In some cases, growth and mental retardation may also be seen. Diagnosis depends entirely on the clinical features.¹¹

Differential diagnoses of our case include Goldenhar syndrome,^{3,12} Hanhart syndrome,⁶⁻¹² and OFD syndromes types I-IX.^{6,9,12,13} Goldenhar syndrome, also known as oculo-auricular-vertebral (OAV) syndrome, has unilateral deformity of the external ear, facial asymmetry and vertebral anomalies. It occurs sporadically though autosomal dominant and recessive patterns have both been described in some families. Hanhart syndrome is a spectrum of hypoglossia - hypodactylia and in the severest form aglossia - adactylia. The major findings in this entity is rudimentary tongue and meromelia.^{12,13} Other findings include micrognathia and peromelia. Although, once thought to be an autosomal dominant disorder, non-Mendelian pattern of inheritance has also been described.^{6,7,12}

Orofaciodigital syndrome type I was first described in 1961 by Gorlin *et al.*¹² Significant features are cleft of the jaw and tongue in the area of the lateral incisors and canines, syndactyly, clinodactyly, and brachydactyly. Other findings include small nostrils, lobulated tongue with hamartomas, irregular and asymmetrical cleft of the palate, aberrant hyperplastic oral frenula, and spotty alopecia. In addition, there is severe malformation of the brain. OFD type II is the closest differential to our case. It was described in 1941 by Mohr.^{12,13} The features include polydactyly, syndactyly, brachydactyly, lobulated tongue with papilliform protuberances, angular form of the alveolar process of the mandible, and supernumerary sutures in the skull. Other findings include cleft palate, pectus excavatum and tachycardia.¹² OFD type III was first described in 1971. The features are limb anomalies, mental retardation, lobulated tongue, hamartoma on the tongue, dental anomalies, bifid

uvula, post axial hexadactyly of hands and feet, pectus excavatum, short sternum, kyphosis, hypertelorism and myoclonic jerks.¹² OFD type IV is characterized by lobulated tongue, pseudocleft of the lip, hyperplastic frenula, pre- and post-axial polydactyly of hands and feet, broad nasal root, lingual hamartomata, severe talipes equinovarus, mesomelic limb shortness associated with tibial hypoplasia and severe bilateral deafness.

Morphologically, the closest differential to our case is Mohr syndrome (OFD type II). The features in our case are compared and contrasted with those of Mohr syndrome in Table I which shows that there are features which differentiate our case from Mohr syndrome.

The challenges experienced in the management of the present case included financial constraints of the parents and their unwillingness to attend follow-up clinic. The basic investigations carried out were done from the department's purse. Genetic studies, though considered, were not readily available and these were to be finalized if patient had presented for follow-up. Although from the literature, the diagnosis of oral-facial-digital syndromes is essentially clinical, identifying the specific type necessitates further radiological and genetic studies which our centre lacked at the time the child presented. The rarity of the case coupled with a dearth of literature on children of African origin with OFD is the other reason for this report. In addition, some neglect or outright abuse of this baby could not be completely ruled out as refusal of the parents to bring the child for follow up may just be a manifestation of their disinterest in the baby because of the abnormalities.

Table I
Comparison between Mohr Syndrome (OFD type II) and the Present Case

Features	Mohr Syndrome	Present Case
General appearance	Usually small for gestational age	Appropriate for gestational age
Nose	Broad and depressed nasal bridge	Bilateral outgrowth in the nostrils
Skull bone	Supernumerary sutures	Normal suture lines
Tongue	Tongue cleft, nodular	Severe hypoglossia
Urogenital system	No reported anomalies	Micropenis
Chest	Pectus excavatum	Pectus carinatum
Lip/palate	Midline partial cleft of the lip with occasional cleft of the palate	No cleft in lip; palatal irregularity without cleft.

We suggest the formation of a national or regional centre for genetic studies where dysmorphic babies can be seen, or in the alternative, specimens from such cases can be sent for detailed studies at no or highly subsidized cost.

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