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Improving prenatal detection of congenital hand defects through collaborative goal-directed antenatal care: a case report on symbrachydactyly

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ABSTRACT

Background: Prenatal ultrasonography for the detection of fetal structural anomaly is an important component of antenatal care. During the assessment, proximal limb deformities are readily diagnosed. Distal limb, especially digit, abnormalities, however, may be difficult to detect, particularly if the ultrasonography is performed in the third trimester, and the deformity is unilateral and isolated.
Case: A 24-year-old primigravida booked for antenatal care with a general practitioner had threatened miscarriage at 12 weeks of gestation, and at 34 weeks was referred to an obstetrician for further care and delivery. The growth ultrasonographic examination was normal but at 40 weeks of gestation she developed antepartum haemorrhage of unknown origin. She had a caesarean delivery and a female baby with "rudimentary" left fingers ("isolated symbrachydactyly") was delivered. The parents were counselled and they declined further assessment of the baby by a hand surgeon and a clinical geneticist. At 3 years of age, the baby had normal development and "is using her hand even without fingers," according to the mother.
Conclusion: Collaborative goal-directed antenatal care that involves different categories of healthcare professionals, but particularly a certified sonologist who performs fetal anomaly ultrasonography, is essential for the detection of congenital hand defects. Adequate counselling is essential for the satisfaction of the baby's family.

1. Introduction

Congenital hand defects or differences range from subtle defect to complete absence of the limb but the classification is controversial and determined by dysmorphological concepts [1] with an option to further describe sub-variants [2]. Hypodactyly is unilateral absence or rudimentary bulbous finger(s) without a terminal ectodermal component [3]. This is different from symbrachydactyly, which is usually unilateral and the rudimentary digit nubbin contains any of cartilage, bone and nail plate [4]. However, symbrachydactyly is used as an umbrella term that includes hypodactyly [4]. The classification of the International Federation of Societies for Surgery of the Hand (IFSSH), which is the most popular [5], enlists symbrachydactyly [6] and not hypodactyly. Regardless, congenital hand deformities develop during the 4–8 weeks following fertilization [7]. Symbrachydactyly results from defective axial differentiation or formation of the hand plate and the limb in its entirety [6,8]. The underlying aetiology is usually unknown but a popular hypothesis as proposed by Bavinck and Weaver is dysgenesis of the subclavian artery in what they termed subclavian artery supply disruption sequence (SASDS) [9]. Symbrachydactyly is usually not a component of a genetic abnormality [8] in the same way that many cases of hypodactyly are idiopathic [3]. Symbrachydactyly was first described by Alfred Poland in 1841, and he named the concurrent occurrence of symbrachydactyly and absent/hypoplastic pectoralis major muscle 'Poland syndrome', [8] which is usually sporadic, although a few familial cases have been reported [10]. Congenital upper limb deformity occurs in 5.25 per 10,000 live births while the subset symbrachydactyly is rarer and complicate 0.6 live births per 10,000 [8].

Following childbirth, both the accoucheur and the family members of the baby with congenital hand defect may be surprised and emotional, particularly if the hand defect was not detected or suspected prenatally. Hence prenatal detection of hand deformity is essential through comprehensive antenatal care that includes a fetal anomaly ultrasonography examination at 18–22 weeks of gestation. Unfortunately, in low- and middle-income countries many pregnant women begin antenatal care or get referred to a specialist obstetrician only in the third trimester. This makes it difficult to detect fetal anomalies, particularly hand

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and digit deformities, which are visualized with prenatal ultrasonography in only 20–30% and 4–19% of cases respectively [11]. The failure in detection may arise because distal limb deformities are difficult to detect, particularly in the third trimester, because of fetal attitude (such as the difficulty in visualizing the hand positioned at the back of the trunk) and normal decreases in amniotic fluid volume that occur with increasing gestational age. It is not surprising that the International Society of Ultrasound in Obstetrics and Gynaecology (ISUOG) does not recommend finger counting as a component of routine mid-trimester fetal anomaly ultrasonography [12].

Here, we report a case of symbrachydactyly which was diagnosed at birth subsequent to no fetal anomaly screening in the antenatal period in a primigravida. This is an example of problems encountered in low- and middle-income countries where second-trimester fetal anomaly scans are not part of routine care. We discuss how to improve the prenatal detection of congenital hand differences through collaborative goal-directed antenatal care that involves different cadres of healthcare professionals, but particularly the feto-maternal medicine specialist. The key elements of the initial management of the condition are also discussed.

2. Case Description

A 24-year-old primigravida booked for antenatal care with a general practitioner. She had threatened miscarriage at 12 weeks of gestation. At 34 gestational weeks she was referred to a specialist obstetrician for further care because the same general practitioner did not conduct delivery. The patient had not been exposed to chemicals, nor did she have a family history of congenital anomaly. She was HIV negative, blood group A positive, rapid plasma reagin non-reactive, HBsAg negative and had a rubella IgG of 18.5 IU/L.

At 40 weeks of gestation she developed antepartum haemorrhage of unknown origin and had an uneventful caesarean delivery. She was delivered of a female baby with Apgar scores of 8 and 9 at 1 and 5 min respectively, and birth weight of 3470 g. Immediately after delivery, physical examination of the baby showed a left hand with "rudimentary" fingers with no nail plate (Fig. 1). The parents of the baby were counselled. X-ray of the baby's left hand showed absent carpal bones (which may ossify later), hypoplastic metacarpal bones and no bony component in the "rudimentary" fingers (Fig. 1). The clinical features were consistent with a diagnosis of isolated symbrachydactyly. The paediatrician, clinical psychologist and orthopaedic surgeon reviewed the baby and counselled the family. In the first week postpartum, the parents did not want the hand abnormality to be assessed further. During follow-up counselling, however, the parents did want further assessment of the hand abnormality, and they were referred to a clinical geneticist and a hand surgeon. Subsequently, they did not go for the further assessment because of their personal conviction that the baby was healthy and needed no additional review. Three years later, the child had normal development and the parents were happy that she "is using her hand even without fingers," according to the mother.

3. Discussion

The commonest type of congenital birth defect in Africa is musculoskeletal abnormality [13] and this includes limb defects such as symbrachydactyly. Again, symbrachydactyly may be a part of Poland syndrome, which in a few cases may be familial. Other syndromes that may be associated with symbrachydactyly are Moebius, Adams-Oliver, amniotic band disruption, del 22q11.2 and Turner (45,X) [10]. Additionally, a congenital defect such as Apert syndrome which manifests as syndactyly, acrocephaly and mutation on fibroblast growth factor receptor −2 (FGFR2) is a differential diagnosis of symbrachydactyly [4]. Therefore, fetal anomaly screening is essential for early detection and management. Invasive testing such as amniocentesis offers the opportunity to diagnose associated syndromes and exclude differential diagnosis of symbrachydactyly that has genetic abnormality as a component. Fig. 2 is a flow diagram illustrating collaborative antenatal care to detect fetal anomaly and the initial postpartum management of congenital hand deformity. To improve prenatal detection of congenital hand defects, a high-resolution ultrasound machine with the settings adjusted for optimal image should be used. On ultrasonography at 11 to 13 weeks 6 days of gestation the bony segments of the 4 limbs and well-aligned hands and feet should be identified [14]. At 18–22 weeks, ultrasonography can identify the fetal trunk and locate the humerus and subsequently systematically identify the elbow, ulna and radius, the wrist and the hand [15]. The normal reflex is for the fetus to close and open his/her hands and this makes it challenging to detect hand defects. During the ultrasonography, it should be noted if the fingers are overlapping when the baby clinches his/her hand. Additionally, 3 phalanxes in each finger should be seen with the hand outstretched. Notably, absence of other fetal anomaly makes it difficult to suspect congenital hand defect. This was the situation in the case reported. In the third trimester, the fetus is grown and the structures are poorly visualized, particularly with normal decreases in the volume of amniotic fluid, which contribute to poor acoustic window.

When selective versus routine ultrasonography in early pregnancy (≤24 weeks) are compared [16], the authors prefer the latter. This is because it affords the opportunity of early detection of major fetal anomalies [16], risk categorization and referral of high-risk pregnancies to an appropriate facility/clinician. Undoubtedly, antenatal care improves pregnancy outcomes [17] but to ensure that ultrasonography at 18–22 weeks is performed with a high detection rate for fetal anomaly, collaborative goal-directed antenatal care is required. This involves

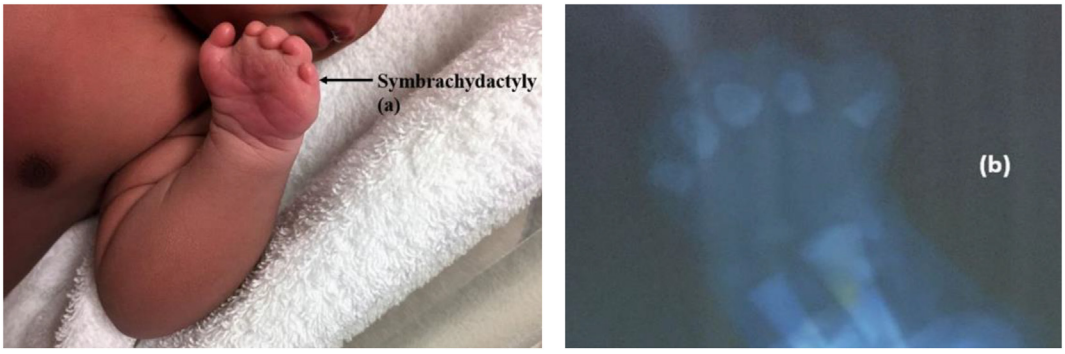

Fig. 1. Symbrachydactyly (a) and X-ray (b) of the left hand at birth.

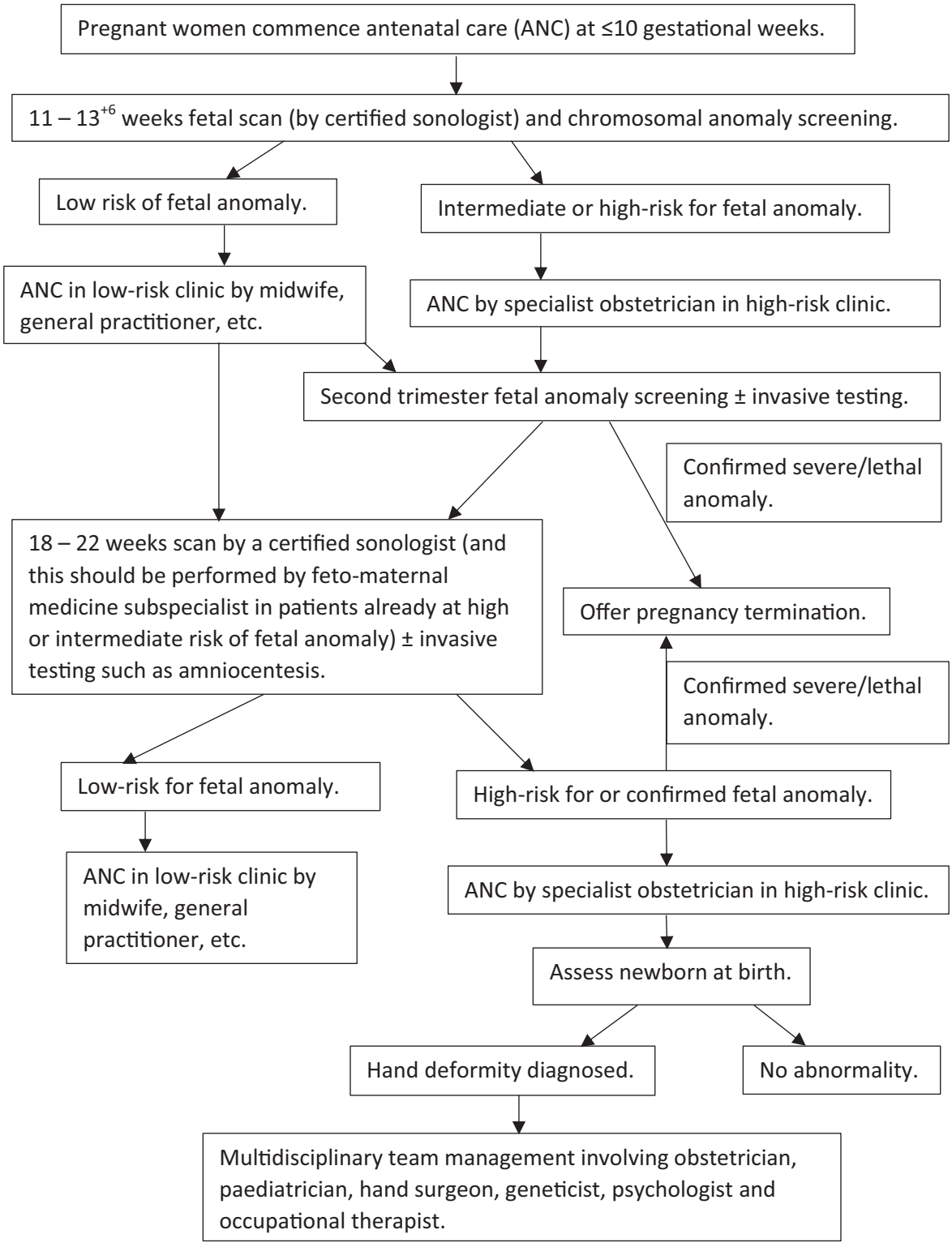

Fig. 2. Collaborative antenatal care to detect fetal anomaly and the initial management of congenital hand deformity.

re-engineering of purposeful well-timed referral of patients among different categories of healthcare professionals. The absence of one-stop antenatal care clinic in many settings makes this model valuable. Goal-directed antenatal care has a high yield for each clinic visit and is preferable to a traditional antenatal care schedule [18]. Understandably, clinicians managing pregnant women should acquire the skills to assess fetal morphology with ultrasonography. In the real world, it is challenging for all clinicians (including general practitioners) managing pregnancy to acquire and maintain all the skills to perform fetal anomaly scans. The following measures are therefore necessary as part of efficient antenatal care: (i) each pregnant woman should be sent for fetal structural anomaly scan at 18–22 weeks of gestation; (ii) promote and

increase the number of training posts in feto-maternal medicine to improve the number of the subspecialists available to provide fetal medicine services; (iii) address the issue of increasing costs of medical indemnity, which have sky-rocketed in a country such as South Africa, which makes it scary for many doctors to undertake ultrasonography assessment of the fetus; (iv) medical practitioners who manage pregnant women who do not have access to a feto-maternal medicine specialist should refer them to an accredited sonographer who can perform structural anomaly ultrasound, and the findings of the sonographer should complement those of the generalist clinician / obstetrician; (v) patients who have an abnormality or who are difficult to assess should be referred directly to a feto-maternal medicine unit. The practice of allowing an accredited sonographer to perform fetal anomaly ultrasonography is already in existence but needs to be strengthened. While this alternative arrangement may be acceptable, feto-maternal medicine specialists are the best group of clinicians to perform fetal structural anomaly sonography but they are scarce and expensive, particularly in low- and middle-income countries.

Antenatal monitoring, delivery and neonatal care as well as the prognosis of identified anomalies deserve consideration when managing congenital musculoskeletal deformity [19]. The parents of a baby who has a congenital anomaly such as symbrachydactyly should be counselled and offered the opportunity for further assessment of the abnormality to improve their participation in care and acceptance of the outcomes. A multidisciplinary team care is essential and should involve: a geneticist to offer pre- and post-test genetic counselling in the antenatal and postnatal periods, depending on when the diagnosis is made; a neonatologist to assess the neonate and exclude other anomalies; a hand or other dedicated surgeon to offer and plan surgical reconstruction; a psychologist to offer psychological support; and an occupational therapist to provide measures to improve hand dexterity. Of note, the unilateral nature of symbrachydactyly influences many parents to delay surgical reconstruction until manifestation of functional deficit in the use of the hands [8]. In the case presented, the baby's parents declined assessment by a geneticist and a hand surgeon, and were satisfied with the management.

4. Conclusion

Congenital distal limb defects are difficult to detect. Collaborative goal-directed antenatal care involving a sonologist certified to perform fetal anomaly ultrasonography will improve the detection of deformities. Effective counselling must be provided to the affected family and they should be offered further expert management.

Contributors

Nnabuike Chibuoke Ngene conceptualized the study and drafted and revised the manuscript.

Laurence Chauke revised the manuscript.

Both authors saw and approved the final version.

Conflict of Interest

The authors declare that they have no conflict of interest regarding the publication of this case report.

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Patient Consent

The patient (mother of the baby) gave written informed consent for the case to be published.

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