

CONGENITAL CLEFT LIP, CLEFT PALATE AND LIP FISTULAE (VAN DER WOUDE SYNDROME) IN TWO NIGERIAN FAMILIES

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تتضمن هذه الدراسة عرض إصابة عائلتين نيجيريتين بوهاد خلقية في الشفة السفلية مترافقة مع شق أحادي أو ثنائي الجانب للشفة والحنك مع فتق أربي .
لم نتطرق إلى تناذر (فان ديروود) كون الدراسات قد بينت شيوع حدوثها لدى الشعب النيجيري .
في الحقيقة تبدي الإصابات التي وردت إلى عيادة المستشفى تغير في موقف الشعب تجاه التشوهات الولادية .
إن معالجة وهاد الشفة إما بالإصلاح الجراحي أو ترك التشوه كما هو عليه .
إن الغاية من الإصلاح الجراحي تأمين الناحية الوظيفية والتجميلية . إن الشذوذات الوجهية والتشوهات المعممة في أجزاء أخرى من الجسم اعتبرت كأعراض معقدة في تناذرين محددين .
إن تناذر الظفرة المأبضية المقترحة من قبل (نحورلين) والمؤلفة من مجموعة من الأعراض المعقدة : ظفرة ما بين الساقين ، التشوهات الأصبعية ، تشوهات الجهاز البولي التناسلي ، وشذوذات الفم .
تتضمن تشوهات الفم ، شق الشفة ، شق قبة الحنك ، وهاد الشفة السفلية ، والكفاف السنخي داخل الفم ، التحام شبه خيطي لأجفان العين قد ورد ذكره في حالات أخرى .
شذوذات الوجه المتضمنة شق الشفة وشق قبة الحنك قد ورد ذكرهم أيضاً في تناذر فقدان الأصابع واللسان المتضمن غياب اللسان وعدم نمو الأصابع ، إضافة إلى حدوث القدم المدببة ، تعدد الأصابع ، وصفر اللسان ، يمكن أن تترافق بتناذر فقدان الأصابع واللسان وإن كن نادرة الحدوث .
اعتبر (شورر) أن ٧٥٪ من حالات شق الشفة تعزى إلى الوراثة المتنحية بينما ٢٥٪ تعزى إلى الوراثة المتسلطة . وجد (سيرفينكا وزملاؤه) بأن ٧,٠٪ من حالات الشفة المشقوقة تكون مترافقة بوهاد الشفة وتعزى الإصابة إلى الانتقال الوراثي لها .
الشذوذات الصبغية مثل فقدان الصبغيات يمكن أن تترافق مع شق الشفة وشق قبة الحنك والاضطرابات الوجهية القحفية الأخرى (نحورلين وزملاؤه) بينما أن نسبة شيوع وهاد الشفة تتراوح بين و
وتعطي الحالات المعروضة أدلة كافية للانتشار العائلي بدرجات متفاوتة .
ذكر (فان درود) تناذر آخر معقد ومعروف منذ حوالي ١٢٠ عاماً يتألف من شق الشفة وشق قبة الحنك والناسور الخلقى للشفة السفلية وتشوهات الأطراف ، وينتقل كصفة وراثية مسيطرة .
تم عرض حالتين لعائلتين مصابتين بشق الشفة وشق قبة الحنك وهاد الشفة في آن واحد ، وذلك تأكيداً للانتقال الوراثي لهذه الاضطرابات وإشارة إلى أن عادة التخلص من الأطفال الرضع بالقتل في نيجيريا قد انخفضت بشكل كبير وهذا يعود إلى التغيرات الإيجابية التي كان من نتائجها حضور عدد أكبر من حالات الشذوذات الوجهية إلى عيادات المستشفى .

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الحالة الأولى:

كانت بعمر سبعة عشر عاماً بين الفحص السريري سلامة الفتاة بشكل عام مع وجود شقة الشفة ثنائي الجانب مع شق قبة الخنك إضافة لذلك تبين وجود وهاد الشفة ثنائية الجانب ومتناظرة بعمق ٥, ٢ سم.

الحالة الثانية:

طفل رضيع بعمر سبعة أشهر الشكوى تتضمن عدم قدرته على مص ثدي أمه وخروج الطعام من الأنف والفم. أظهر الفحص السريري شق أيسر أحادي الجانب في الشفة وقبة الخنك مع وهاد شفوية ثنائية الجانب بعمق ٥, ٠ سم أظهر الفحص السريري العام سلامة الطفل مع وجود فتق أربي أيسر.

Two Nigerian families with congenital lower lip pits in association with unilateral or bilateral cleft lip and palate and inguinal hernia are described. Van der Woude Syndrome was not reported among the Nigerian populace in the literature. The fact that these cases presented in the hospital clinic shows a change in the attitude of the populace towards birth defects, it can be reasonably concluded that an era of infanticide is closed or drawing to an end. Treatment modalities for lip pits are surgical repair or leaving it alone as in one of our cases. Clefts of the lip and palate are surgically repaired to achieve the objectives of proper speech, function and aesthetics.

Introduction

Facial anomalies and generalized abnormalities in other parts of the body have been reported as symptom complexes in two well-defined syndromes.¹

The name Popliteal Pterygium Syndrome was suggested by Gorlin *et al* for the symptom complex that consists of intercrural pterygium, digital anomalies, genito-urinary and oral anomalies. The oral anomalies include cleft lip, cleft palate, lower lip pits and intraoral alveolar webbing. Reported cases have also shown filiform adhesion of the eyelids.²

Facial anomalies, including cleft lip and cleft palate, are also reported in the Aglossia-Adactylia Syndrome in which the tongue is absent with failure of the development of the digits.³ Club foot, polydactylism, spina bifida and microglossia, though rare, can also be associated with Aglossia-Adactylia Syndrome.

Schroder⁴ considered that 75% of the cleft lip cases are due to recessive inheritance while 25% are due to dominant inheritance. Cervenka *et al*⁵ found that 0.7% of the patients with cleft lip and palate had concomitant lip pits. They also attributed genetic transmission to this disorder.

Chromosomal aberrations in the form of deletions may be associated with cleft lip and palate and other craniofacial disorders.⁶ Gorlin *et*

al reported the frequency of lip pits in the population to be between 1:75,000 and 1:100,000. From the reported cases in the literature, there is abundant evidence of familial occurrence of variable expressivity. Van der Woude¹⁰ reported another syndrome complex consisting of cleft lip, cleft palate, congenital fistulae of the lower lip and anomalies of the extremities. The same author ascribes this syndrome, which has been recognized for over 120 years, being transmitted as an autosomal dominant trait. Pits of the upper lip have been reported but it appears unlikely that they are related to this syndrome.¹¹

Although over 250 cases of Van der Woude Syndrome have been reported, more are being reported because of its variable expressivity.^{8,12,13} However, we are not aware of cases being reported in the Nigerian population. Two families in which cleft lip, cleft palate and lip pits occurred together are reported to reinforce the genetic transmission of these disorders and highlights the fact that infanticide has been greatly reduced in Nigeria. This is a result of positive changes in cultural attitudes making it possible for more cases of these facial anomalies to attend the hospital clinics.

Case 1

A 17-year-old girl was referred from a district

hospital to the Oral Surgery Clinic of the Dental School Hospital of the University of Benin with a bilateral cleft lip.

Examination revealed a healthy looking girl with bilateral cleft lip and palate and a prominent prolabium. There was also a symmetrical bilateral lip pits which measured 2.5 cm in depth [Fig. 1]. The fistulous tract drained some mucus when expressed. No other abnormality was detected.



Figure 1. Bilateral cleft lip and palate with a symmetrical bilateral lip pits.

The family tree [Fig. 2] was interesting. Mother had seven children (5 girls and 2 boys). Only four are alive (3 girls and 1 boy). All girls had the same defects including the labial pits. It was not present in the parents or the boys. Three of the children died in infancy. The family pedigree could not be traced back because of lack of records and cultural attitudes.

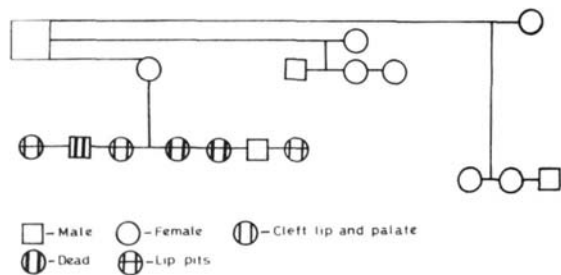


Figure 2. Family tree of Case 1.

The father married two other wives and none of the children have cleft or lip pits. The patient and the elder sisters have had the repair of their clefts done. However, they were not interested in the repair of the labial pits.

Case 2

A 7-month-old baby boy was brought to the Oral Surgery Clinic of the Dental School and Hospital of the University of Benin with complaints of inability to suck the mother's breast and regurgitation of food from the nose and mouth.

Examination revealed a left complete unilateral cleft of the lip and palate with bilateral labial pits [Fig. 3]. The pits measured 0.5 cm in depth. Systemic examination revealed a healthy child with an associated (L) inguinal hernia.



Figure 3. Left complete unilateral cleft lip (cleft repaired) with bilateral labial pits.

While taking the history from the father, it was observed that he also had bilateral symmetrical labial pits [Fig. 4]. There was no history of cleft lip and palate or labial pits in his family or that of his wife. The child was the first and only child of the family at the time of presentation.

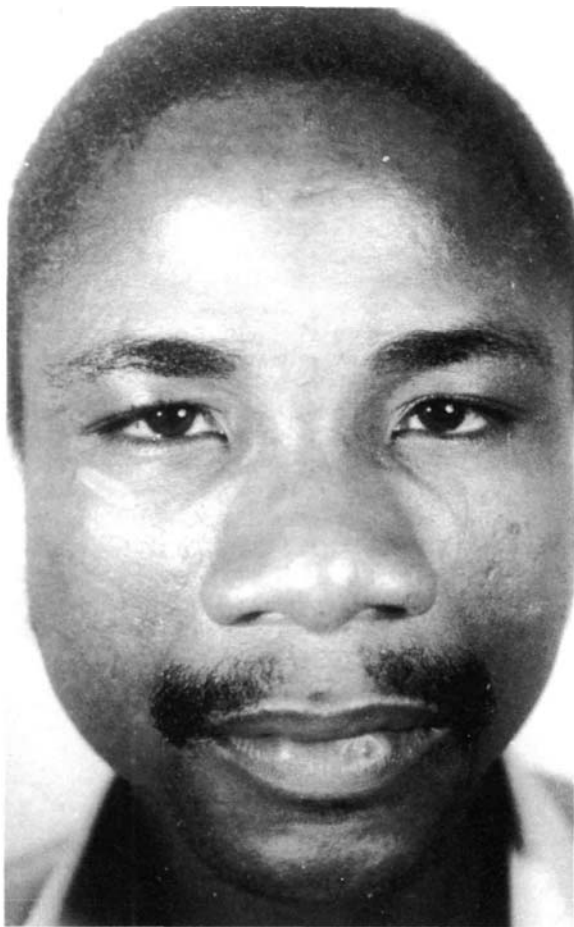


Figure 4. Father of child (Case II) with lip pits.

Discussion

Asymmetrically and bilaterally placed lip pits may be associated with bilateral cleft lip and palate (Case 1) or unilateral cleft of the lip and palate (Case 2).² Lip pits placed bilaterally and asymmetrically may occur without concomitant palatal and labial clefts as in the father of Case 2. The openings of the fistulae were round and mucus material were drained when expressed as in Case 1, but was of no embarrassment to her.

Other oral manifestations of this syndrome in addition to cleft lip, cleft palate and lip fistula have been reported.^{1,34} It is interesting to note that Case 2 has an associated left inguinal hernia conforming the variable expressivity of the syndrome.¹ The fact that these dentofacial anomalies are new, attending the hospital clinics may be due to

improved educational attainment, positive changes in cultural attitudes towards birth defects and acceptance of medical explanation for their disability. Good results from treated facial clefts also serve as convincing evidences that these conditions could be repaired.

Ord and Sowray¹⁴ and our present experiences¹⁵ show that infanticide, which Strauss¹⁶ feels remain a practice in Nigeria, is no longer practiced. The difficulty which Oluwasanmi and Adekunle¹⁷ encountered in obtaining a family history of congenital anomalies as a result of denials may now have been reduced because of positive changes in cultural attitudes towards these anomalies.

Some studies^{17,14} have adequately covered the aetiology of lip pits and facial clefts. Khan *et al*¹⁸ and Svristava and Bang¹⁹ have speculated that Van der Woude Syndrome may, in fact, be a mild variant of popliteal pterygium with variable expressivity and incomplete penetrance. The pattern of inheritance of Van der Woude Syndrome in the girls of the family of Case 1 could not be explained since tracing the family pedigree back was not possible. Chromosomal deletion may be a possibility but we could not do chromosomal examination due to lack of facilities. The disorder's occurrence only in girls of this family could be explained when family pedigree and chromosomal examination are available. The risk of inheriting a cleft from an affected parent is estimated at about 20% and the risk of inheriting lip pits at about 25%.¹²

Although these conditions are rare, they may become rarer with attention being focused on genetic counselling.

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