Case Report

Dysmorphism in One of a Set of Male Twins; Could They have been Identical Twins? A Diagnostic Dilemma

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Abstract

Twinning occurs worldwide, but Nigerian women of the southwest extraction record one of the highest rates in the world. Among the notable risk factors for fraternal twinning is advanced maternal age, which is also an independent risk factor for Down syndrome. Even as morphological characteristics can easily be applied to distinguish identical from fraternal twins, in cases of an associated dysmorphism, in any member of the couplet, it becomes a difficult tool to use, as has been observed in the case of a set of twins who were of the same sex, blood group, hemoglobin genotype, and shared the same placenta.

Keywords: Advance maternal age, down syndrome, dysmorphism, identical twins

Introduction

Twinning is common among Nigerian women, especially of the south western extraction,[1] and it has been related to the consumption of a species of yam containing phytoestrogen, which stimulates ovulation.[2] Other factors associated with twinning are advanced maternal age[3] and with the advent of the assisted reproductive technique, the twining rate has increased.[4] Even as these hold true for fraternal twins, which is the most common; this is not the case for identical twins. Twinning in Nigeria occurs with a frequency of 40.2 per 1000 deliveries, which is among the highest rates in the world.[5] The mechanisms involved in multiple pregnancies also predispose couplets to various forms of congenital malformation.

Increasing maternal age also independently heightens the risk of Down syndrome to 1:1600 in those less than 25 years, rising to 1:350 at 35 years, and 1:40 at the age of 43 years.[6] Despite this, substantial cases of Down syndrome occur in children of young Nigerian mothers.[7]

Case Report

A seven-month-old boy, delivered as twins, was noticed to have recurrent difficulty in breathing since the age of five months. This was associated with fever, but no history of cyanosis or altered consciousness. He was born to a 46-year-old mother and a 50-year-old father in a non-consanguineous marriage setting. This was the first delivery of the mother. They were both male, shared the same placenta, and were of the same blood group.

On examination, he had a low set of ears, upward slanting of the palpebral fissure, prominent bilateral epicanthic folds, and a broad nasal base [Figure 1], with umbilical hernia. He appeared dysmorphic when compared to the other couplet [Figure 2]. The cardiac examination revealed tachycardia, with an apex beat at the sixth left intercostal space at the anterior axillary line. He also had a pansystolic murmur at the left lower sternal margin radiating upward.

Full blood count, electrolyte, and a urea and thyroid function test were not remarkable; however, a chest x-ray showed cardiomegaly [Figure 3] with pulmonary plethora. An electrocardiogram showed evidence of biventricular hypertrophy, while an echocardiogram revealed a peri-membranous ventricular septal defect [Figure 4], with left-to-right shunting; and karyotyping revealed Trisomy 21 (47 × Y + 21). The diagnosis of Down syndrome in discordant twins was made; however, the possibility of them having been identical by virtue of their having shared the same placenta, same blood group, and sex was also entertained.

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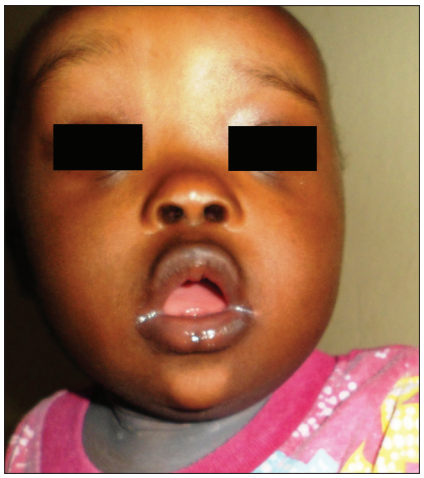

Figure 1: Upward slanting of the palpebral fissure, prominent bilateral epicanthic folds, and broad nasal base

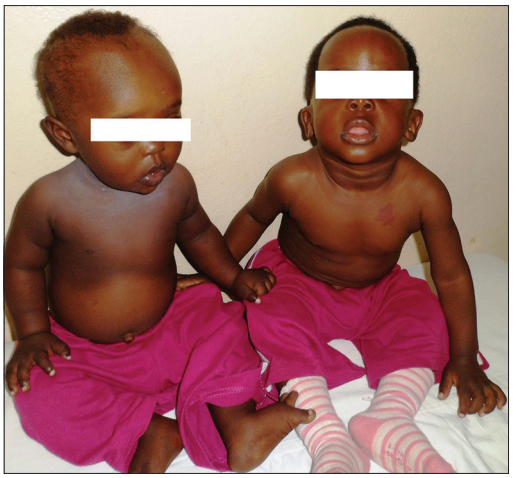

Figure 2: Morphological discordance in the pair

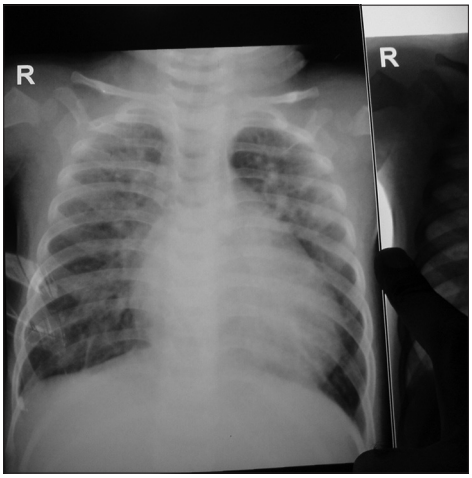

Figure 3: Chest X-ray showing cardiomegaly

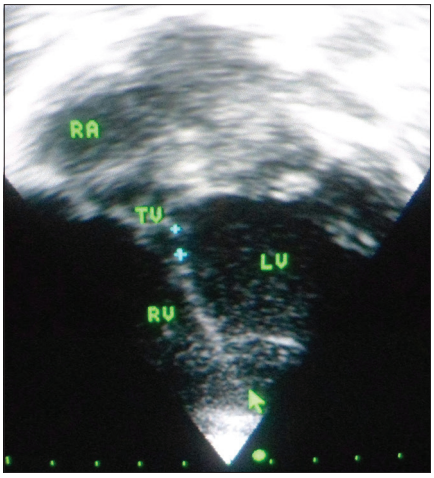

Figure 4: Echocardiogram showing a sub-aortic ventricular septal defect

Discussion

The index case had clinical features that were consistent with the Down syndrome, which was further confirmed by karyotyping. It was reported that 11.8% of 244 twin pairs had Down syndrome and it was estimated that identical twins with Down syndrome occurred at the rate of one to two per million pregnancies, while in non-identical twins it occurred at the rate of 14-15 per million.[8,9]

Monozygotic twins are genetically identical and they are always of the same sex, unless mutation occurs in one of the pair during early fetal development. However identical twins can be remarkably different morphologically; they may be of a different size at birth, as it is seen in the twin–twin transfusion syndrome and may also differ both intellectually and behaviorally especially if raised differently. This is mostly due to the effect of environmental modifications, which may also influence gene expression; furthermore, increasing epigenetic differences with increasing age may manifest with morphological and intellectual differences. Some of these environmental factors may be intrauterine; therefore, it not surprising that identical twins do not have the same fingerprints. This is because of the differential uterine wall impact, resulting in differences in their appearance. Therefore, identical twins may occasionally appear non-identical.[10]

Other factors include random deactivation of X chromosomes in female monozygotic twins and aneuploidy, affecting the phenotypic characteristics of one of the identical pair.[11]

The index cases were both male and of the same blood group and hemoglobin genotype, which is seen in identical twins. Furthermore they had the same placenta, but different amniotic sacs, however, they lacked similar facial characteristics, which was due to the dysmorphism in the first of the couplet.

The pregnancy had a heightened risk of Down syndrome because of advanced maternal age, but why it only occurred in the first of the pair is not clear. The plausible explanation could be due to aneuploidy occurring in one of the pair after separation of the zygote. Furthermore each twin could have

had an independent risk for Down syndrome if they were non-identical; therefore, the observed similar blood group and hemoglobin genotype being coincident and proximity of their placenta could have resulted in its fusion making it appear single. However a detailed DNA analysis would have determined their exact zygosity, but this is not routinely available in everyday clinical practice.

Conclusion

Down syndrome in a member of a set of twins makes determining the zygosity difficult, as was observed in this Nigerian family and its exact mechanism is still not completely understood.

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