CASE REPORT

Down syndrome in one of non-identical Nigerian Twins: A case report.

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ABSTRACT

Background: *Down syndrome (DS) or Trisomy 21 is the most frequent and best known Trisomy in humans. Mothers under 25 years of age are known to have the average risk of a DS pregnancy of 1:1600, rising to 1:350 at 35 and 1:40 at age 43. Twining in DS occurs at a rate of 1.2% of pregnancies with only 1/6th of both of the pairs having Trisomy 21.*

Method: *A 4 week old male, second twin, was admitted in the University of Port Harcourt Teaching Hospital (UPTH) due to fast breathing from birth, cough and poor weight gain. He was very pale, (haematocrit 18%), dyspnoeic and had mongoloid features. There were coarse crepitations in lung fields, a systolic murmur and an enlarged liver. The diagnoses were Down syndrome, bronchopneumonia and congenital heart disease with failure. The mother was aged 22years and the twin sister was normal.*

Result: *He was promptly treated with oxygen, diuretics, and antibiotics but died within three hours of admission before blood transfusion could be offered.*

Conclusion: *Down syndrome in twin pregnancy is very rare and, to our knowledge none has been reported in Nigeria. That this was a product of first pregnancy in a lady as young as 22years makes an interesting reading.*

Key Words: *Down syndrome: Discordant Nigerian Twin; Young mother Paper accepted for publication 12th September 2006*

INTRODUCTION

Down Syndrome is the most frequent and best known of the trisomies in humans[1]. It was first described by the London physician, John L Down in 1886 but its cause was not known until 1959 when Lejeune and Turpin showed that the individual with DS carried 47 chromosomes, the extra being chromosome 21[1].

Maternal age is the only well-recognized association of Down Syndrome.[2] Mothers below 25 years have an average risk of a DS pregnancy of about 1:1600. The risk rises to about 1:340 at 35, 1:40 at age 43,[2] 1:20 at 47 and 1:12 at 49 years[3]. However, it has been suggested that the risk of an affected birth does not increase after about age 43. In a review of the National Down Syndrome Cytogenetic Register (NDSCR) in England, 44% of diagnoses were at birth[4]. Of all pregnancies with prenatal diagnosis of Trisomy 21, 94% of parents decided to terminate the pregnancy, 5% were live born and 1% were stillbirths or neonatal deaths[4].

Twining occurs at a rate of 1.2% in pregnancies in the NDSCR, and Trisomy 21 in both of a set of twins occurs in one sixth of these.

The rare combination of Downs Syndrome in one of a set of twins of a young mother is being reported.

CASE REPORT:

Baby O. J. a 4 week old male, 2nd of a set of twins, (the 1st a female) was rushed into the UPTH Children emergency ward with a 4-week history of fast breathing, 2 week of cough and poor weight gain (compared to twin1) and poor suck of four days duration.

The infants were delivered in a private clinic by spontaneous vertex at term to a booked 22-year old primiparous undergraduate. A mid-trimester ultrasound scan showed twin gestation but pregnancy was however uneventful. Patient was delivered 10minutes after the first twin. Birth weight was 2.5kg while sibling weighed 2.7kg. He was exclusively breastfed until one week prior to presentation when SMA gold was introduced, which he took poorly. Immunization was up to date. Father is 35 years of age. There was no family history of consanguinity and no family history of multiple pregnancies. The mother did not use fertility-enhancing drugs.

On examination, he was pale, with an axillary temperature of 38.6°C and in severe respiratory distress with flaring alae nasi, supraclavicular, intercostal and subcostal recessions. He had generalized hypotonia,(Fig 1) with flat face, low set ears (Fig II), upwardly slanting palpebral features and wide epicanthic folds. He had a single palmar crease (Simian) with clinodactyly of the fifth finger. There was a wide gap (sandal gap) between the first and second toes. (Fig 111)

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FIG 1: showing discordant twins: Note frog-like posture of twin with Down's Syndrome .

Fig11: Affected twin with DS. Note flat face and low set ears

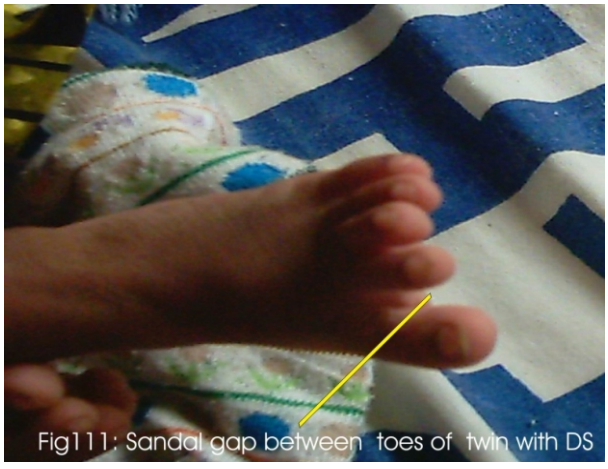

Fig111: Sandal gap between toes of twin with DS

Weight was 2.8kged (compared to twin who weighed 4.6kg). He was tachypnoiec with a respiratory rate of 82/minute and had bilateral basal crepitations. He had a tachycardia with a heart rate of 190/minute with normal first and second heart sounds, and a pansystolic murmur which was maximal at the left lower sternal border. There was a tender and enlarged liver which was 6cm below the right costal margin. He was conscious but lethargic with generalized hypotonia.

Emergency resuscitative measures instituted included nursing in a cardiac position, oxygen by nasal catheter, and intravenous frusemide at 1mg/kg. Urgent PCV done was 18%. Blood was sent for grouping and crossmatching. However, the child died within three hours of admission and before blood was made available for transfusion.

Discussion

The clinical features of Down Syndrome first described by John Langdon Down in 1886 and can be variable[5]. The infant in this report had more than five features, whereas the presence of two or more suggests a possible diagnosis of Down Syndrome and the need for chromosomal studies[5].

The incidence of DS is estimated to be more than twice as high in all conceptions as it is among live births[1].More than half of the trisomy 21 conceptions abort early in pregnancy. The occurrence of Trisomy 21 increases with advancing maternal age. The increased risk of trisomy 21 in women over 35years is an indication to offer these women amniocentesis or chorionic villi sampling and chromosomal analysis as a way to detect fetal Down Syndrome[1]. In women under 35years of age, maternal á-fetoprotein concentration is lower, unconjugated estriol decreases and human chorionic gonadotropins increases in the presence of Down Syndrome in the foetus. The combination of these is effective for prenatal screening. It is reported that if amniocentesis is offered to women 35 years and above, about 20% can be diagnosed prenatally. If low serum á-fetoprotein screening is done in patients under 35 years of age, 20-30% of cases of DS can be diagnosed antenatally.[5] Our patient's mother was 22yrs, a primiparous and had no indication for prenatal diagnosis.

If Down Syndrome is suspected in a mother with twins, amniocentesis is not a reliable test because it relies on the fact that babies with DS have higher levels of human chorionic gonadotropins (hcG) and lower á-fetoprotein (AFP) than normal babies. If one of the twins has Down Syndrome and the other is normal, the lower levels of AFP from the one with DS will be masked by the normal levels from the other[6]. Thus a Nuchal Translucency (NT) Scan is the best screening test for Down Syndrome in twin pregnancy[6]. None of these options of diagnosis is available in our centre and our patient had no indication however to do them.

Fifty four percent of babies with trisomy 21 are said to be males. In the discordant twin pairs, 49% are males and where both are affected the male sex accounts for 57%.[4] In this report, the affected child is a male.

Whereas no documentation on the seasonal variation has been made in our environment, variation in season or clustering of DS birth in time or area have been identified in developed countries.[7]

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