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Original Research

Prevalence and diagnostic dilemma of chromosomal abnormalities in the Niger Delta area of Nigeria; Is prenatal diagnosis worthwhile?

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Abstract

Background: Early detection of increasing numbers of cases of chromosomal abnormalities (aneuploides) at the Rivers State University Teaching Hospital (RSUTH) in the Niger Delta will enhance appropriate counseling of patients and early termination of the affected pregnancies. The study aimed to ascertain the prevalence of aneuploides at the RSUTH and to determine the necessity for early prenatal diagnosis in the Niger Delta.

Methodology: This was a prospective cross-sectional study carried out over a three-year period (01/01/2018 – 01/01/2021) at the RSUTH, Nigeria. Newborn babies delivered at 28 weeks and above were assessed at birth for the phenotypes of aneuploidy and associated birth defects. A convenient sampling method was used to recruit all the babies with chromosomal abnormalities and their mothers. Data including that of socio-demographic, obstetric characteristics, and the fetuses were taken and analyzed using Statistical Package for Social Sciences Version 23 (SPSS version 23). Quantitative variables were summarized using means and standard deviation while qualitative variables were expressed as frequencies and proportions.

Results: The total number of babies that were delivered by the 5868 participants in the study was 6078, out of which 10 cases of aneuploides were identified – 3 cases of trisomy 18 and 7 cases of Trisomy 21. The prevalence of chromosomal abnormalities at birth at the RSUTH was 0.165% of the total births, 1:2000 and 1:654 for T18 and T21 respectively. 1:654 mothers had babies with chromosomal abnormalities, 1:2000 and 1:833 for T18 and T21 respectively.

Conclusion: The prevalence of chromosomal abnormalities at birth at the RSUTH of 0.165% represented a gross underestimation because the diagnosis was based on the outward phenotypical appearance of the neonates and it was made not from babies delivered at 20 weeks and above as practiced in Europe and other countries, but from 28 weeks. Prenatal diagnosis was therefore highly recommended in the Niger Delta.

Keywords: Prevalence; Chromosomal Abnormalities; Prenatal Diagnosis; Tertiary Health Facility; Niger Delta; Nigeria.

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Quick Response Code:

Introduction

Some of the known chromosomal abnormalities are Edward syndrome or Trisomy 18 (T18), Down syndrome or Trisomy 21 (T21), Patau syndrome or Trisomy 13 (T13), Turner's syndrome (45OX) and sex chromosome abnormalities. T18 is categorized into full trisomy 18 (about 95% of cases), T18 mosaicism (about 5% of cases), and Translocation T18 (very rare). Mosaic trisomy 18 occurs when both a trisomy 18 and a normal cell line are present in the same individual. The clinical phenotype varies depending on the level of mosaicism. [1] T18 is the second most common autosomal trisomy after trisomy 21. The incidence of T18 in the first trimester was 1 in 400, but due to high spontaneous loss due to cardiac, renal, and CNS defects, the birth prevalence was 1 in 6500. [2,3] Trisomy 18 has no racial predilection. Approximately 80% of cases occur in females. [4,5]

T21 is by far the most common and best-known chromosomal disorder in humans and the most common cause of intellectual disability. [6-10] It is categorized into 3 cytogenetic variants, namely T21 resulting from non-disjunction during meiosis in one of the parents (94% of cases) with 95% of the cases maternal, chromosomal translocation (3.3%) and mosaicism (2.4%). This occurrence is correlated with advanced maternal and paternal age. Translocation occurs when genetic material from chromosome 21 becomes attached to another, like chromosome 14, 13, 15, or 22. The male-to-female ratio is approximately 1.15:1 in newborns with T18 due to meiotic disjunction. 50% of female patients with trisomy 21 are fertile, and these females have up to a 50% chance of having a live child who also has trisomy 21. On the other hand, men with Down syndrome are usually infertile, except for those with mosaicism. There is no racial predilection for Down syndrome.

Prenatal diagnosis includes screening for and diagnosis of aneuploids. Most of the screening tests are done in the first trimester of pregnancy. The screening methods and their associated detection rates (DR) for a false positive result of 5% are as follows: Maternal age (MA)(DR 30%), MA, and maternal serum biochemistry at 15–18 weeks (DR 50–70%), MA and fetal NT and maternal serum free b-hCG and PAPP-A at 11–13+6 wks also called the triple test (DR 85–90%), MA and fetal NT and fetal nasal bone (NB) at 11–13+6 wks (DR 90%), MA and fetal NT and NB and maternal serum free b-hCG and PAPP-A at 11–13+6 wks (DR 95%).[11] Those at high risk are offered CVS or amniocentesis. Presently, in most of the developed world, the triple test and cell-free DNA test, also called non-invasive prenatal diagnosis (NIPT) (DR almost 100%) are offered. Different combinations of screening modalities involving biochemical tests, ultrasound screening, and NIPT are also offered.

Screening for chromosomal abnormalities is also performed at 19-22 weeks of pregnancy by using the identified birth defects (minor and major markers) that are associated with them. [12,13] The presence of the markers modifies the already achieved risk in the first trimester.[12] When an abnormality is detected a thorough check for the other features of the associated chromosomal defect is carried out. Trisomy 21: Nasal hypoplasia, nuchal fold thickness, cardiac defects, duodenal atresia and echogenic bowel, mild hydronephrosis, shortening of the femur, sandal gap and clinodactyly or mid-phalanx hypoplasia of the fifth finger. [12,13]

Associated birth defects with T18 are abnormalities of the head and spine, facial, neck, heart (more than 90% of cases), lungs, diaphragmatic hernia, GI abnormalities, Genitourinary anomalies in 9% of cases, skeletal abnormalities namely shortening of the limbs, radial aplasia, typically overlapping digits, with the second and fifth fingers overlapping the third and fourth fingers, respectively (89% of cases), talipes or rocker bottom feet,[14] fetal growth restriction (29%) and oligohydramnios/polyhydramnios in 12% of cases, Trisomy 13: Holoprosencephaly, microcephaly, facial abnormalities, cardiac abnormalities, enlarged and echogenic kidneys, exomphalos and postaxial polydactyly.

On physical examination at birth, patients with trisomy 21 have characteristic craniofacial findings, such as a flat occiput and a flattened facial appearance, small brachycephalic head, epicanthic folds, upward-slanting palpebral fissures, brushfield spots, lateral nasal bridge, small nose, and small mouth, protruding tongue, small and dysplastic ears, generous nuchal skin. Abnormalities of the hands and fingers, including joint hyperextensibility or hyper-flexibility, sandal gaps and neuromuscular hypotonia, etc. may be present. [15-20] Other presentations are diastasis recti, dry skin, and congenital heart defects.

At birth, the dysmorphic features for T18 are as outlined above, and also the following: eye abnormalities, short nose with upturned nares, choanal atresia, narrow palatal arch, ear abnormalities, hypotonia followed by hypertonia, jitteriness, apnea, seizures, Gastro-intestinal abnormalities, diastasis recti, male external and female internal and external genital anomalies, thymic, thyroid and adrenal hypoplasia, Except in those where NIFT has been done, diagnosis of aneuploidy is normally confirmed in the postnatal period with conventional cytogenetic and fluorescence in situ hybridization (FISH) studies.

In Nigeria, prenatal diagnosis and screening for birth defects are not offered universally as practiced in most European and North American countries. Chromosomal abnormalities are often diagnosed shortly after birth by recognizing dysmorphic features and distinctive phenotypes. This study aimed to ascertain the prevalence of chromosomal abnormalities in babies of parturients at the Rivers State University Teaching Hospital (RSUTH) and to determine the need for the introduction of prenatal diagnosis in the hospital and in the Niger Delta area and Nigeria at large.

Methodology

Study Area -The study was carried out in the labor ward and the Special Care Baby Unit (SCBU) of the Rivers State University Teaching Hospital (RSUTH). RSUTH is a tertiary health facility located in Port Harcourt which is the capital of Rivers State (one of the 36 States in Nigeria).
Study design: The study was of a prospective cross-sectional design carried out over a three-year period between 01/01/2018 and 01/01/2020.
Inclusion Criteria: The inclusion criteria were all pregnant women who delivered at the RSUTH during the study period and their babies.
Exclusion Criteria: Women who did not consent to the study.
Sampling technique: The non-probability, consecutive sampling method was employed to recruit participants into the study.
Study process – All babies that were born from 28 completed weeks of pregnancy and above were assessed at birth on the labour ward and later by the paediatricians in the SCBU for dysmorphic features and distinctive phenotypes associated with T21, T18, and T13. All the babies with chromosomal abnormalities were recruited into the study after their parents had given informed consent. Data on the socio-demographic and obstetric characteristics of mothers and the time that elapsed between delivery and diagnosis were taken on a questionnaire.

Statistical analysis - Data was analysed using Statistical Package for Social Sciences Version 23 (SPSS version 23). Quantitative variables were summarized using means and standard deviation while qualitative variables were expressed as frequencies and proportions.

Ethical Consideration

The study was carried out in compliance with the international ethical guidelines for biomedical research involving human subjects. Ethical approval was obtained from the RSUTH ethics committee. Informed consents were obtained from all the women who were enrolled in the study. All the information that was collected from individual patients was available for clinical use and for research purposes. Privacy rules were maintained, and confidentiality was observed at all levels of dealing with patients' data.

Results

The deliveries and the number of women whose babies had chromosomal abnormalities are shown in Table 1 below while the breakdown of the socio-demographic characteristics of the mothers is shown in Table 2.

Table 1: Pattern of deliveries at the RSUTH from 01/01/2018 to 01/01/2022

Pattern of deliveries	**Number (%)**
Singletons	5681
Twins,	166 (2.83%)
Triplets	19 (0.32%)
Quadruplets.	2 (0.034%)
Total number of deliveries	5868
The total number of babies	6078.
Total number of Women with Trisomy babies	10

Table 2: Socio-demographic characteristics of mothers of babies with chromosomal abnormalities

	T21	**T18**	**Total**
Total Number	7	3	10
Variables			
Maternal age			
35 years and more	5 (50.00%)	0 (0.00%)	5 (50.00%)
Less than 35 years	2 (20.00%)	3 (30.00%)	5 (50.00%)
Mean age of the women with Trisomies			34±4.428,
Maternal education			
Secondary	3 (30.00%)	0 (0.00%)	3 (30.00%)
Tertiary	4 (40.00%)	3 (30.00%)	7 (70.00%)
Marital status			
Married	7 (70.0%)	3 (30.0%)	10 (100.00%)
Not married	0 (0.00%)	0 (0.00%)	(0.00%)
Maternal occupation			
Hairstylist	1 (10.00%)	0 (0.00%)	1 (10.00%)
Nurse	0 (0.00%)	1 (10.00%)	1 (10.00%)
Public Servant	1 (10.00%)	0 (0.00%)	1 (10.00%)
Teacher	2 (20.00%)	2 (20.00%)	4 (40.00%)
Trader	3 (30.00%)	0 (0.00%)	3 (30.00%)

In total, there were 7 babies with T21 from 6 mothers and 3 with T18 from three mothers (Table 3), totaling 10 babies from 9 mothers. Six of the trisomic babies (five with T211 and one with T18) were diagnosed within 1 day of delivery, while one baby with T21 was diagnosed within 2 days, another baby with T21 and two with T18 were diagnosed within 3-8 days of delivery. The diagnosis was based on the recognized dysmorphic features and the distinctive phenotype of the conditions. Further neonatal clinical and maternal obstetric variables are shown in Table 3.

Table 3: Clinical variables of babies and Obstetric variables of mothers of babies with chromosomal abnormalities.

Variables	T21 (%)	T18 (%)	Total
Days before diagnosis			
Day 1	5(83.33)	1 (16.67)	6
Day 2	1 (100.00)	0 (0.00)	1
Day 3-8	1 (33.33)	2 (66.67)	3
The mean Age of the Neonate at final diagnosis			2.67±0.71 days
Total number of affected foetuses	7 (70.00)	3 (30.00)	10
Number of babies			
Singleton	5 (62.50)	3 (37.50)	8
Twins	1 (100.00)	0 (100.00)	2
Parity			
0	1(100.00)	0 (0.00)	1
1	3 (60.00)	2(40.00)	5
2	0 (0.00)	1(100.00)	1
3	1(100.00)	0 (0.00)	1
5	1(100.00)	0 (0.00)	1
Presentation			
Cephalic	4 (57.14)	3 (42.86)	7
Breech	3 (100.00) including I of the twins	0(0.00)	3
Mode of delivery of the babies			
Spontaneous Vaginal Delivery	2 (50.00)	2 (50.00)	4
Caesarean section	5 (80.00) including the twins	1 (20.00)	6
Birth weight			
2.5 kg and more	4 (80)	1(20)	5
Less than 2.5 kg	3 (60)	2 (40)	5
The mean Neonatal birth weight			2.78±2.77 Kg
Gestational age at birth			
37 and more weeks	2 (66.67)	1 (33.33)	3
32-36 weeks	4 (66.67)	2 (33.33)	6
The mean Gestational age at birth			37.89±2.89 weeks
Sex			
Male	4(100.00)	0 (0.00)	4
Female	3 (50.00)	3 (50.00)	6

The perinatal outcome of babies with chromosomal abnormalities is shown in Table 4. Out of the 10 babies that were delivered, six (five with T21 and one with T18) were discharged by the pediatricians while three (two with T21 and one with T18) were discharged against medical advice. One baby with T18 died in hospital.

Table 4: Perinatal outcomes of babies with chromosomal abnormalities.

Outcome	T21	T18	Total
Discharged	5(83.33)	1 (16.67)	6
Died	0 (0.00)	1(100.00)	1
Discharged against medical advice	2 (66.67)	1 (33.33)	3

Nine women out of the total 5868 had 10 trisomic babies; the total number of delivered babies was 6078. Aneuploidy occurred in 0.165% of all the delivered babies. That was equivalent to 1.65:1000 or 1:606 babies at birth. Also, 0.153 of the total women who delivered during the study period had babies with chromosomal abnormalities. That was equivalent to 1.53:1000 or 1:654 women that had life births.
Further data on the outcome of the pregnancies and prevalence of chromosomal abnormalities are shown in Table 5.

Table 5: Pregnancy outcome and prevalence of the Chromosomal abnormalities within the study period

Variable	Prevalence
Singletons	5681 (96.86%)
Twins	166 (2.83%) = 332 babies
Triplets	19 (0.32%) = 57 babies
Quadruplets.	2 (0.034%) = 8 babies
Total number of women	5868 (5681+166+19+2)
The total number of babies	6078 (5681+332+57+8) .
Total number of Women with Trisomy babies	9
Isolated T18	1
T18 in association with intersex and spinal bifida	2
Total number of T18 babies	3
Prevalence of T18 babies	3/6078 x 100 = 0.05% of life births = 5 in 10000 live births
	That is equivalent to 1:2000 live births.
	3/5868 x 100 = 0.05% of the mothers – 5 in 10000 Mothers
	That is equivalent to 1:2000 Mothers.
Trisomy 21	7 (Including 1 set of twins)
Prevalence of T21	7/6078 x 100 = 0.12% of live births = 12 in 10000 live births.
	That is equivalent to 1:833 live births.
	7/5868 x100 = 0.12% of women = 12 in 10000 Maternities.
	That is equivalent to 1:833 Maternities.
Total number of Trisomy's	10
Prevalence of Aneuploidies	0.165% (10/6078 x 100) of the delivered babies That is equivalent to 1.65:1000 (0.165x1000/100) or 1:606 (1/1.65x1000) babies at birth.
Prevalence of Mothers with Tr. babies	0.153% (9/5868 x 100) of the maternities had babies with chromosomal abnormalities. That is equivalent to 1.53:1000 women (0.153x1000/100) or 1:654 (1/1.53x1000) women that had life births.

Discussion

The study was prompted by the increasing number of chromosomal abnormalities at birth in the Rivers State in the Niger Delta area of Nigeria. [21] Out of the six mothers that had babies with T21 one was less than 35 years of age while 5 (83.33%) of them were more than 35 years. Paradoxically, the 3 women who had T18 babies were all below 35 years of age; that was contrary to the popular belief that T18 just like T21 occurred more in older women than younger ones. [22] The women that had T21 babies delivered at earlier gestation1 ages - 4(66.67%) at 32-36 weeks of pregnancy than at later gestations - 2(33.337%) at 37 weeks and more. The findings were in tandem with the popular belief that the risk for T21 increases with maternal age and decreases with gestation. [22, 23] The prevalence of trisomy 21 at 12 and 16 weeks of gestation was higher than that at 40 weeks; they were 30% and 21%, respectively.[23]

The incidence of T18 in the first trimester was 1 in 400, but due to high spontaneous loss, the birth prevalence was 1 in 6500. Approximately 95% of conceptuses with trisomy 18 die as embryos or fetuses. [2, 3, 24] For live-born infants with trisomy 18, the estimated probability of survival to age 1 month was 38.6%, and to age 1 year was 8.4%.[7] Long-term survivals up to age 27 years have been reported. [2, 3, 24} In trisomy's 18, 13, and Turner syndrome, the rate of fetal death between 12 and 40 weeks is about 80%.[25]

In T21, the two sexes are affected roughly equally. The male-to-female ratio is slightly higher (approximately 1.5:1) in newborns with free T21. The same principle was applicable to the findings in the present study where 4(57, 14%) of the 7 neonates that had T21 were boys while 3(42.86%) were girls. In the case of T18, the 3 babies were girls, contrary to the evidence that approximately 80% of trisomy18 cases occur in females. [26,27] Generally, maternal age, gestational age at birth, and neonatal sex impact on the prevalence of trisomies at birth.

In the present study, 5868 women had babies during the study period. 9 (0.153%) of them had babies with chromosomal abnormalities. That was equivalent to 1.53:1000 (0.153x1000/100) or 1:654 (1/1.53x1000) women that had life births. The total number of babies that were delivered during the study period was 6078. Regarding the prevalence of trisomies among the neonates, 0.165% (10/6078 x 100) of the delivered babies had trisomies. That was equivalent to 1.65:1000 (0.165x1000/100) or 1:606 (1/1.65x1000) babies at birth.

The first case of T21 in Nigeria was described in a case report in 1964.[28] An incidence of 1 in 865 live births was reported in a study in Ibadan, Western Nigeria.[29] This is like the findings in this research. In another study, cytogenetic analysis in 386 patients showed that 369 women (95.5%) had regular trisomy 21, 2.5% - translocation trisomy 21, 6(1.5%) were mosaics while the remaining 2(0.5%) cases were classified as miscellaneous.[30] A high incidence of cases among young mothers was recorded. Nuchal translucency (NT) scan was first carried out in Nigeria in 2010.[31] Another study correlated it with VSD and NTD but not with aneupliody.[32]

1:2000 Mothers had babies with T18 while 1:833 of them had babies with T21. The prevalence of T18 was 1:2000 live births while that of T21 was 1:833 live births. The above statistics are far less than the prevalence of chromosomal abnormalities that were diagnosed in England in 2020 for trisomy conditions: Down's syndrome 1 in 377 births, Edwards' syndrome 1 in 1,352 births, Patau's syndrome 1 in 3,707 births. [33] In the present study, there was no statistical data on trisomy 13. The prevalence of T18 of 5:10000 (1.2000) was comparable with the finding in another study which showed it to be 5.85:10000. [34] It was also comparable with the data from the congenital anomaly registers in Europe in 2012 which showed the prevalence of T18 to be 5.9:10000. [35]

The prevalence of T18 of 1:2000 live births was far less than that in the United States of 1 in 6500 at birth.[2, 3] The prevalence of T21 of 12:10000 (1:833 or 0.12%) in the present study was also far less than 23.40:10000 and 23:10000 in the Chinese and European studies respectively.[34,35] In the European study, the cases were taken from 16 population-based registries in 11 European countries, and the diagnosis was made prenatally or before 1 year of age between 2000 and 2006. They included live births, fetal deaths from 20 weeks gestation, and terminations of pregnancy for fetal anomaly.[35]

It is important to note that the present study only concentrated on the prevalence of T18 and T21 at birth. In the other studies that were compared with the present one, data were collected from 20 weeks of pregnancy and included both live births, fetal deaths, and those pregnancies that were terminated due to fetal abnormalities, and in some cases, data were collected up to one year of age of the babies. It is a known fact that the prevalence of trisomy 21 at 12 and 16 weeks of gestation was higher than that at 40 weeks; they were 30% and 21%, respectively.[23] In the same vein, the prevalence of T21 at birth should be far less than that at lower gestations. Furthermore, approximately 95% of conceptuses with trisomy 18 die as embryos or fetuses.[2, 3, 24] In trisomies 18 and 13 and Turner syndrome, the rate of fetal death between 12 and 40 weeks is about 80%.[25] Therefore, the prevalence of T18 and T21 that were achieved in the present study, using neonatal phenotype did not represent the true prevalence of the conditions in the Niger Delta. They should be far higher than that and even higher than the figures that were obtained in China, Europe, and the United States.

In the present study, neonatal phenotype alone was used as a diagnostic tool for aneuploidy because prenatal diagnosis is rarely practiced in Nigeria. Ultrasound- and biochemistry-based tests and non-invasive prenatal diagnostic tests based on maternal cell-free DNA tests are not universally done in Nigeria. However, there are sporadic cases of amniocentesis, karyotyping, and PCR tests. Furthermore, second-trimester ultrasound scans for major and soft markers of aneuploidy are not done. Availability of those prenatal screening and diagnostic tests will drastically increase the prevalence of chromosomal abnormalities in the Niger Delta, including T21, T18, T13, Turner's syndrome, and sex chromosome abnormalities.

Recommendation

It is recommended that prenatal diagnosis should be introduced in the Niger Delta and Nigeria at large. At the initial stage of the introduction of prenatal diagnosis, biochemical tests, namely pregnancy-associated protein-A (PAPP-A) and HCG assay may not be available. What can be offered at the Rivers State University Teaching Hospital are the following screening tests with their associated detection rates (DR) for a false positive result of 5%: Maternal age (MA)(DR 30%), MA and fetal NT and fetal nasal bone (NB) at 11–13+6 wks (DR 90).[36, 37] The detection rate can be increased by adding more tests namely fetal heart rate, fetal tricuspid regurgitation and ductus venosus.[38] Those who are at high risk are then offered invasive diagnostic procedures CVS or amniocentesis which are feasible in Nigeria today.

Even if the screening cannot be made universally available, elderly gravidae (from 35 years of age), women who had babies with chromosomal abnormalities, and those who in the index pregnancy have babies with ultrasound phenotypes of aneuploidy should be screened for the condition. In women who had a previous pregnancy with trisomy 21, the risk of recurrence in the subsequent pregnancy is 0.75% higher than the maternal and gestational age-related risk for the condition at the time of testing. [25] This occurs because less than 5% of couples with a previously affected pregnancy have parental mosaicism or a genetic defect that interferes with the normal process of disjunction. So, in this group, the risk of recurrence is increased substantially. In most couples (more than 95%), the risk of recurrence is not actually increased.[30]

Limitations: This study used only the neonatal phenotype in the diagnosis of the chromosomal anomalies and so the prevalence found may be less than the actual prevalence. Nonetheless, it shows the presence of the chromosomal abnormalities amongst our people and the need for proper screening to identify more cases. Though this is a single centre study the result may be generalisable because the Rivers State University Teaching Hospital is in the metropolitan city of Port Harcourt and receives referrals from other neighbouring communities and States.

Conclusion

The prevalence of chromosomal abnormalities (T18 and T21) at birth at the RSUTH was remarkable and eye-opening. Nevertheless, these figures may represent a gross underestimation of the true prevalence because they were taken at birth, (pregnancies from 20 weeks were not included as practiced in Europe) and diagnosis was based solely on the outward phenotypical appearance of the neonates. Prenatal diagnosis is highly recommended in the Niger Delta and Nigeria.

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