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Phocomelia in an HIV infected baby: Case report

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Introduction

Phocomelia refers to a birth defect in which the hands and feet are attached to abbreviated arms and legs. The term comes from phoco (meaning 'seal') and melia (meaning 'limb'), to indicate a developmental abnormality in which the limb is like a seal's flipper. There has been an explosion of knowledge about child development in past decade or so, and it is hard to remember that it was only about 50 years ago that the discovery was made that the fetus is vulnerable to exposures. The phocomelia epidemic resulting from use of thalidomide by pregnant women was an early but dramatic example of the ability of chemicals to traverse the placenta and damage the fetus. More than one system can be susceptible and different pathologies may occur depending upon the dose and timing of exposure.

Case Report

Baby EEU was a 4-hour old female baby delivered via spontaneous vertex delivery to a booked 35 year old Gravida five para five (two males and three females) woman at a gestational age (GA) of 40weeks at the University of Calabar Teaching Hospital (UCTH) on 10th October 2012. The Apgar scores at birth were six and seven at one and five minutes respectively. The baby showed fair activity at birth and needed minimal resuscitation. Pregnancy was booked at UCTH at 17 weeks gestational age but she defaulted and was not seen again until 30 weeks of gestation. The mother had the following laboratory profile: blood group O Rhesus D positive, haemoglobin genotype AA, HIV seropositive. She however refused antiretroviral drugs (ARV) treatment. At 30weeks gestation she had a febrile illness and severe diarrhoea which necessitated hospitalization for five days. Thereafter she accepted ARVs (no Efavirenz) but defaulted to follow

up. Her medical record did not show any record of CD4 count prior to commencement of drugs at the hospital. The medical history did not indicate any rash during the first trimester, drugs or herbal ingestion apart from routine hematinics. There was no history of alcohol ingestion or smoking during pregnancy. Her highest level of education was JSS 2 and she sells cooked food.

The father, a 40-year-old army rating with SS3 as the highest level of education, was found to be seroreactive to HIV five months after wife's diagnosis.

The baby girl weighed 2.2kg and had a number of congenital abnormalities: the right upper limb was amputated at the proximal arm; there was a rudimentary left arm with three digits and no forearm. In addition there were right mandibular hypoplasia and asymmetry of the jaw and mild pectus carinatum. Her length and Occipito frontal circumference (OFC) were 47cm and 33cm respectively. The lower limbs were normal and no other abnormalities were detected. The baby was seropositive to HIV using ELISA and DNA PCR. Other viral studies like rubella cytomegalovirus serology could not be done because of lack of necessary facilities to conduct such investigations. The working diagnosis was HIV embryopathy with gross skeletal abnormalities in a preterm low birth weight baby. The baby's CD4 count was 1200cc/mm², and her E/U/Cr, and haemogram were essentially normal. Baby was managed at the Special Care Baby Unit (SCBU), was discharged after two weeks and followed up at the pediatric outpatient clinic. The social works department of the University of Calabar Teaching hospital was also involved in the management of this baby.

Fig 1 and 2 shows the limb (phocomelia) and jaw abnormalities (right mandibular hypoplasia and micrognathia)

Fig 1



Fig 2



Out of the 839,521 births in the five geographic regions, 10,844 birth defects were recorded with a prevalence of 129 per 10,000 births.

In Uganda⁶, of 754 new born babies delivered in Mulago Hospital, Kampala, 33 babies (4.4%) were diagnosed with external birth defects. Limb defects accounted for 45.7% while cleft lip and palate made up 14.2% of all defects. Also common were central nervous system defects (8.5%), omphalocele and spina bifida, 5.8% each, other anomalies, together, constituted 20.0%.

In Ile Ife, Nigeria, a total of 624 neonates were examined, 43 (6.9%) of whom had external birth defects. Musculoskeletal malformations observed in 21 (3.5%) newborns constituted the largest number of birth defects⁷.

Discussion

Birth defects are structural, behavioral, functional and/or metabolic disorders that a baby may have at birth. The known factors are grouped into genetic/chromosomal disorders accounting for 15% of birth defects, while environmental factors and twinning accounted for 10% and 0.5-1% of the birth defects respectively¹.

Approximately 2-3% of births are associated with major congenital defects². Although the cause of birth defects is still not known in 40-60% of cases, it has been established that several factors can put an embryo at risk. There is, however, a variable frequency in different populations, ranging from 1.07% in Japan³ to 4.3% in Taiwan⁴. It is also important to note that regional variations occur in the prevalence of specific birth defects. Rankin *et al*⁵ described trends in total and live birth prevalence of birth defects, and the regional differences in prevalence of the defects among five British regions.

Many investigators have described HIV embryopathy as a condition characterized by craniofacial defects, including microcephaly, hypertelorism, box-like head, and saddle nose, long palpebral fissures with blue sclera, a triangular philtrum, and patulous lips⁶. However, many investigators have since questioned the significance of these observations. Such researchers indicate that there is lack of evidence for characteristic craniofacial malformations in infants who acquired HIV infection from their mother before, during, or shortly after birth (i.e., perinatally)³.

In the case report under review, we found that the baby presented with some craniofacial disorders like right mandibular hypoplasia and asymmetry of the jaw in addition to limb abnormalities. The authors decided to report this case to raise the awareness of clinicians as well as ask if any causal relationship can exist with HIV.

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