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Presumed 13q deletion syndrome in a Nigerian child: A Case Report

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Introduction: 13q deletion syndrome is a rare genetic disease caused by the deletion of some or all of the long arm of chromosome 13. Patients with 13qdeletion syndrome are at risk of retinoblastoma when the RB1 gene, located in the chromosome band 13q14 is deleted proximally. Other features include Dandy Walker mal-formation, cerebellar hypoplasia, and agenesis of corpus callosum.[1] To the best of our knowledge, there has been no reported case in Nigerian children. We report a presumed case of 13q deletion syndrome.

Case Report: We present a 3-year-old boy first seen in our facility when he was 11 months old with poor vision in both eyes noted at the age of 5 months. He is a product of twin pregnancy achieved by in vitro fertilisation and carried by a surrogate mother. He was delivered at 26 weeks gestation with birth weight of 1.07kg, and had insertion of a ventriculoperitoneal shunt at 3 months for hydrocephalus with absence seizures. General Examination revealed plagiocephaly while systemic examination was unremarkable. Ocular examinationrevealed roving eye movements. The right eye had a central cornea opacity with no view of the fundus, left eye revealed normal anterior segment with a whitish retrolental mass.

Ocular ultrasound and Magnetic resonance imaging (MRI) of the Brain and orbits revealed features suggestive of bilateral retinoblastoma (Groups E and D in the right and left eyes respectively). His caregivers were counselled and the he subsequently underwentright enucleation (retinoblastoma was confirmed by histology [Figure 1]) followed by 6 cycles of chemotherapy (Carboplatin, Etoposide and Vincristine). He was followed up in the clinic while having chemotherapy. He defaulted follow up visits after the chemotherapy and later presented with cornealopacity in the left eye which hindered examination of the posterior segment. He subsequently had repeat MRI of the brain and orbits as well as ocular ultrasound scan, about 12 months after the initial MRI, which revealed a well defined left intraocular globular shaped retina lesion measuring 16mm by 9.5mm by 15mm (Figure 2). The child had repeat course of 6 cycles of chemotherapy (Carboplatin, Etoposide and Vincristine) and is currently being followed up in the paediatric ophthalmology and oncology units.

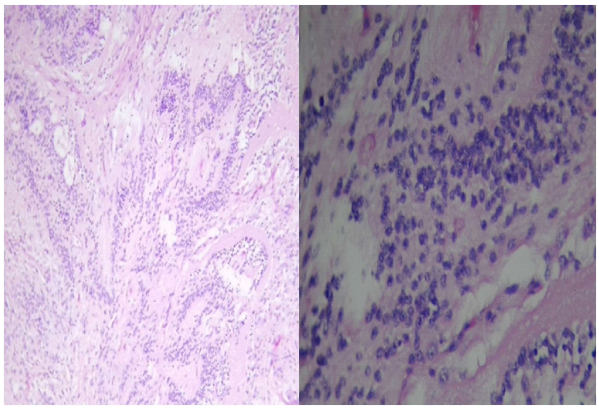

Figure 1: *Histology slides showing necrotic and haemorrhagic invasive tumour growing in sheets, trabeculae, and solid nest of small round blue cells having hyperchromatic nuclei and scant cytoplasm, scattered within the tumor cells are Homer-Wright and Flexner-Wintersteiner rossettes.*

Discussion: Chromosome 13q deletion syndrome was first described by Allderdice after studying two paediatric patients in 1969[2]. The first patient affected by the syndrome including retinoblastoma was reported in 1983[2]. Clinical characteristics, phenotypic description and severity depend on the size of the deleted region

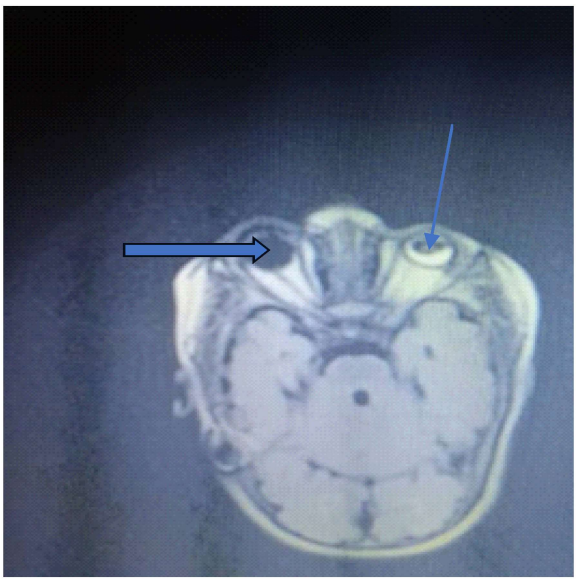

Figure 2: *T2 Weighted FLAIR MRI Brain and orbits showing enucleated right eye with prosthesis (thick blue arrow) and a well-defined left intraocular retinal based globular shaped mass (thin blue arrow).*

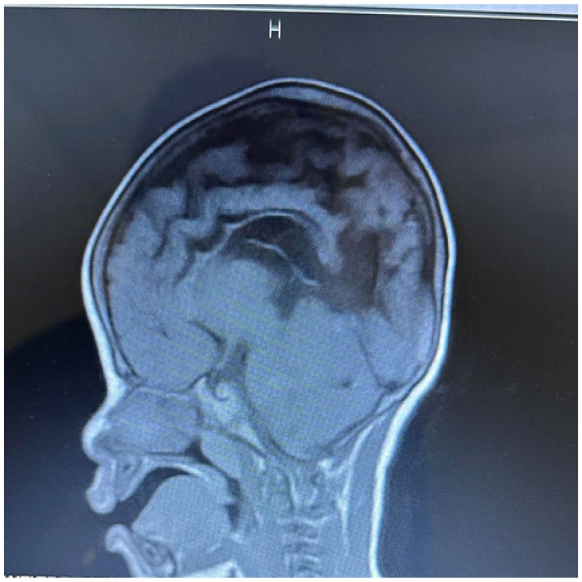

Figure 3: *Absence or Agenesis of the corpus callosum.*

and the location on the chromosome[1,3]. Clinical features of the syndrome vary widely among patients.Growth retardation, microcephaly, facial dysmorphism, cardiac malformations, and mental retardation constitute the major features of the syndrome[4]. The syndrome is currently divided into 3 groups: Group 1 are deletions proximal to 13q32, Group 2 includes 13q 32 while Group 3 are those distal to 13q 32[5]. Patients are at risk of retinoblastoma when the RB1 gene, located on the chromosome proximally is deleted[1]. Distal deletion is associated with severe mental retardation but there no major malformations and growth deficiency[1,6]. Our patient presented with retinoblastoma, phagiocephaly, absence seizures, hydrocephalus and agenesis of corpus callosum (Figure 3) similar to features reported by Wang *et al*[1], Allderdice *et al*[7], and Ballarati *et al*[8]. Their cases also had genitourinary abnormalities and cardiac defects which were absent in our patient[1]. Patients with 13 q deletion are reported to present with corneal opacity[3]. Our patient had this feature which was not found in the cases reported by Wang *et al*[1] and Gargallo *et al*[2]. The presence of retinoblastoma, agenesis of corpus callosum, little or no mental retardation and other anomalies in our patient is strongly suggestive of proximal deletion and is likely Group 1 type of 13q deletion syndrome. The features of Group 1 (proximal deletion cases) include growth deficiency, mild mental retardation, and minor anomalies. Patients with Group 1 disease can also present with microcephaly, hypertelorism, a depressed nasal bridge, simian creases, mild hypotonia and retinoblastoma[5].

Diagnosis is made on the basis of a thorough medical history including family medical history, physical examination, laboratory, radiological studies such as brain scan and chromosomal studies.Management of 13q deletion syndrome usually involves a multidisciplinary team of specialists.

In conclusion, although 13q deletion syndrome is a rare condition, efforts should be made to look out for features of the syndrome in patients who present with hydrocephalus and retinoblastoma. Improved access to genetic analysis may contribute to better outcomes for 13q deletion syndrome in Nigerian children.

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Corneal diameter of preterm babies seen at a tertiary hospital in North-Central Nigeria

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Background: Corneal Diameter (CD) is an essential clinical diagnostic and monitoring tool in the practice of paediatric ophthalmology. Measuring corneal diameter is useful in the diagnosis of various corneal diseases. Normative data on our local population of preterm babies is desirable. The aim of this study was to determine the mean corneal diameter in preterm neonates and to correlate it with their birth parameters.
Methods: Healthy preterm babies delivered before 37 weeks of gestational age were consecutively recruited into the cross sectional study after obtaining necessary institutional approvals from Ethics and Research Committee of the Hospital. The babies were laid in supine position to take the measurement of the white-to-white vertical and the horizontal corneal diameters at recruitment in their first week of life and at 40th week post conceptional age (PCA) using the Castroviejo caliper. Statistical analysis was performed using IBM-SPSS-25.
Results: Ninety-six preterm neonates were recruited, 46(47.9%) were males with a male to female ratio of 1:1.1. The range and mean± standard deviation (SD) of birth parameters including the gestational age, birth weight, birth length and occipito-frontal circumference were 26-36 weeks (32.8±2.29), 0.75-2.73kg (1.71±0.41), 32.0-48.0cm (40.66±3.08) and 23.0-34.0cm (30.28±2.52), respectively. In the first week of life, the mean±SD horizontal corneal diameter in the right and left cornea was 8.93±0.51mm and 8.85±0.51mm, respectively (p=0.293). Similarly, the mean±SD vertical diameter in both right and left corneas was 8.85±0.51mm. The values had increased by 40th week PCA. Figure 1 shows the distribution of horizontal corneal diameter in each eye across the different gestational ages at first week of life. There was a strong positive correlation between the corneal diameter and the birth parameters in the preterm babies. (Table I)