Wolf-Hirschhorn Syndrome with Buphthalmos

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Abstract: Wolf-Hirschhorn syndrome is a 4p-deletion syndrome in which there is reduced growth, abnormal phenotype, developmental delay, intellectual disability and seizures. We are hereby reporting a case which showed typical phenotypic facial features at birth. The case is being presented as this baby had Buphthalmos which is rare.

INTRODUCTION

Wolf-Hirschhorn syndrome is a microdeletion of band 4p16.3. The frequency is estimated to be 1 case per 50,000 births. It is more common in females. Ocular abnormalities found in this syndrome are hypertelorism, strabismus, refractive errors, epicanthal folds, proptosis, downsloping palpebral fissures, microphthalmos, microcornea, iris coloboma, optic nerve coloboma, ocular cysts, ptosis, glaucoma and nystagmus [1]. It is characterized by other facial features like microphthalmia, micrognathia, a short philtrum, small philtrum, small upper lip, a downturned mouth, poorly formed ears with pits or tags (Greek warrior helmet) appearance. Slow growth begins even before birth in this condition. Hypotonia is a marked feature of this condition in the newborn period. Developmental delay especially in verbal communication and language skills are seen later in life [2]. Additional features of Wolf-Hirschhorn syndrome are skin, skeletal and dental abnormalities, cleft palate and cleft lip. It is associated with abnormalities of the eyes, heart, genitourinary tract and brain. The common heart defects include atrial and ventricular septal defects but hypoplastic left heart syndrome was also reported associated with this condition [3]. Nearly 35% of patients die during the first year of life, due to congenital heart defects. Diaphragmatic defects are also reported with this condition [4].

Case Report: The baby was born of a non consanguineous marriage to a 19 yr old lady by Caesarean section. The baby was a term male baby with birth weight of 2.1 Kg (intrauterine growth retardation). There was no history of exposure to radiation, drug intake or fever during pregnancy. Abnormal facial features noted were prominent Glabella, broad bridge of the nose, small philtrum, small upper lip, a downturned mouth, micrognathia and low set ears.

Ophthalmic examination revealed prominent eye balls, downward slanting palpebral fissure and Hypertelorism. Inferonasal coloboma was noted in the left eye. Under sedation horizontal corneal diameter was measured to be 14 mm in both the eyes (megalocornea) using Vernier calipers. Intraocular pressure was measured by using hand held Perkins Tonometer and was found to 29.0mmHg in the right eye and 26.0 mmHg in the left eye (Normal being 11-20mmHg). Dilated retinoscopy revealed myopic astigmatism in both eyes. Dilated fundus examination showed increased cup-disc ratio of 0.6 with normal vessels and macula.

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Systemic examination and skeletal survey did not show any abnormality except for hypotonia of all four limbs. There was no similar history in the family.

**DISCUSSION**

Our case had many features of Wolf-Hirschhorn syndrome like hypertelorism, downward slanting palpebral fissure, inferonasal iris coloboma. The salient difference found in this case from previously reported cases was the presence of megalocornea and congenital glaucoma. Increased intraocular pressure in newborn stretches the sclera and cornea which explains the megalocornea rather than a micro cornea usually seen in this syndrome.

This baby had intrauterine growth retardation which is also characteristic of the syndrome.

Microdeletion involves WHSC1, LETM1 and MSX1 genes [5]. It is believed that loss of the WHSC2gene is associated with many of the characteristic facial features of Wolf-Hirschhorn syndrome and developmental delay. A microdeletion of band 4p16.3, can be detected only by molecular probes. Apart from 5-13% of patients showing a translocation responsible for the syndrome the rest is novo deletions, usually on the paternal chromosome 4, or de novo translocations in 1.6%.

To detect the deletion, techniques like micro satellite analysis, fluorescence in situ hybridization (FISH) is indicated. Fluorescence in situ hybridisation of the patient and his parents is done to exclude translocations. Recurrence risk is low in de novo deletions and translocations, but is remarkably increased in familial translocations. Prenatal diagnosis is possible again by FISH [5].

Early diagnosis by identification of facial features and ophthalmic evaluation will help in managing the learning difficulties.

**REFERENCES**