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Neonatal Progeroid Syndrome

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Abstract

Neonatal Progeroid Syndrome also Known as Weidemann Rautenstrauch Syndrome is an extremely rare genetic disorder characterised by a senile appearing face (old man look) & Pseudohydrocephalus at birth. We describe a patient showing clinical features of Neonatal Progeroid Syndrome.

Keywords: Neonatal Progeroid syndrome; Weidemann Rotenstrauch syndrome; Pseudohydrocephalus; Senile appearing face.

Introduction

Neonatal Progeroid Syndrome is a rare autosomal recessive genetic disorder observed in various ethnic and racial groups. The specific underlying defect responsible for the disorder remains unknown. More than 30 cases are reported till date & 2 cases are reported from India.[1,2]

Neonatal Progeroid Syndrome is characterized by an aged appearance at birth (old man look) delayed growth before and after birth (prenatal and postnatal growth retardation), and deficiency or absence of the layer of fat under the skin (subcutaneous lipoatrophy).[3] It is anticipated that most individuals with Neonatal Progeroid Syndrome have decreased life expectancy. We describe a patient showing clinical features of Neonatal Progeroid Syndrome.

Case Report

A male child born out of second degree consanguineous marriage was delivered to a 22 years primigravida mother by vertex vaginal delivery, post dated by 6 days, cried weakly after birth. Antenatal Ultra sound was suggestive of severe oligohydramnios with Intra Uterine Growth Retardation. Baby was Small for gestational age (SGA) with Birth weight 1.3 Kg, Head circumference 31 cm and length 45 cm at birth. The baby was admitted in NICU for Very Low Birth Weight, dysmorphic features and feeding difficulty.

On general examination baby had an old man look with decreased subcutaneous fat [Figure 2], craniofacial disproportion which gave a pseudohydrocephalic appearance with a triangular face, a large mouth with protruded tongue, sparse hair on scalp,

Figure 1: Pseudohydrocephalic Appearance with Triangular Face

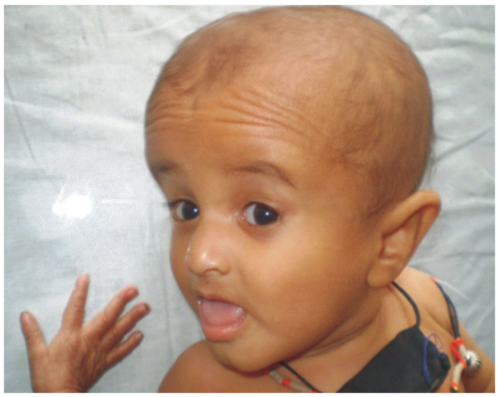
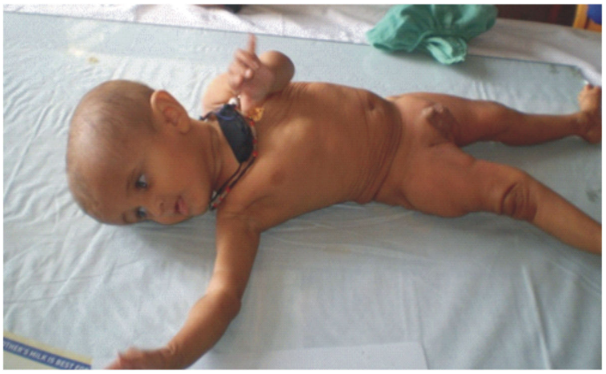


Figure 2: Old Man Look with Decreased Subcutaneous Fat



prominent veins, wide open anterior [Figure 1] & posterior Fontanelle, low set ears,difficulty in establishing feeds, inguinal hernia. Congenital dislocation of hip bilaterally as Barlow & Ortalani test positive. Investigation suggestive of normal lipid profile, USG hip suggestive of Bilateral Hip Dysplasia. Skeletal survey didnot reveal any other abnormality. Karyotype was normal 46XY.

On follow up at 9 months baby has delay in Motor, Language & social developmental milestones. The baby is failure to thrive with Weight, length, Head Circumference below

Table 1: Clinical Features in Neonatal Progeroid Syndrome and Comparison of Features with Previously Reported Indian Cases

S No.	Cinical Feature	JP Narayanan *et al* [1]	Meenu Pandey *et al* [2]	Present case report
1	Antenatal Ultrasound	NR	Oligohydramnios	Severe Olgohydramnios
2	Maturity(Gestational age)	36 weeks	33 weeks	Term
3	Birth Weight	1.5 Kg	910 grams	1.3Kg
4	Birth Length	43 cm	38 cm	45 cm
5	Head Circumference	34 cm	27 cm	31
6	Sex	Female	Male	Male
7	Consanguinity	+	+	+
8	Old man look	+	+	+
9	Pseudohydrocephalus	+	+	+
10	Wide open sutures	+	+	+
11	Wide open fontanell	+	+	+
12	Sparse Scalp Hair	+	+	+
13	Prominent Scalp veins	+	+	+
14	Hypoplasia facial bones	+	+	+
15	Low set ears	+	+	**NR**
16	Beak Shaped Nose	-	+	**NR**
17	Ocular anomalies	+	+	**NR**
18	Dentition present at birth	**NR**	**NR**	**NR**
19	Slender limbs	+	+	+
20	Hypertonia	+	**NR**	**NR**
21	Cryptochidism	**NR**	**NR**	**-NR**
22	Fat accumulation in buttocks	+	+	+
23	Dysplasia of hips	**NR**	+	+
24	Inguinal Hernia	**NR**	**NR**	+
25	Status on Followup	Died on day three of life	Psychomotor retardation at 24 months age Failure to thrive	Psychomotor Retardation at 9 month age Failure to thrive

NR: Not Reported

3rd percentile by WHO Growth standards. Above findings confirmed a clinical diagnosis of Neonatal Progeroid syndrome. Baby will be followed up furthur for evaluation of growth pattern.

Discussion

The clinical features of our case are similar to those described earlier.

The aetiology of Wiedemann-Rautenstrauch syndrome is still unclear; it has been stated that it is not a malformation syndrome, but a disorder that involves mesenchymal tissue, mainly subcutaneous fat. The major physical features remain unchanged in survivors. Evidence for an autosomal recessive pattern of inheritance has been presented in four reports, with possibilities of prenatal diagnosis by ultrasound. Deficient biosynthesis of decorin has been reported, although this is a non-specific and probably secondary phenomenon.[4]

Table 1 Shows Clinical features in Neonatal Progeroid Syndrome and comparison of features with previously reported Indian cases.

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