Barber Say Syndrome (A New Case Report)

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Abstract

Barber Say syndrome (BSS) is a rare ectodermal dysplasia with neonatal onset characterized by congenital generalized hypertrichosis, atrophic skin, ectropion and macrostomia. A literature review showed less than 20 previously reported cases of Barber Say syndrome. This presentation reports a one day old female with syndrome face, low hairline, coarse face, macrostomia, thin upper lip, bilateral ectropion and hypertelorism, hypertrichosis, senile skin appearance, hypoplastic nipples and one area of mild skin atrophy. These findings are consistent with BSS.

Keywords: *Barber Say syndrome, ectodermal dysplasia, ectropion, hypertrichosis*

Introduction

The ectodermal dysplasias (EDs) are congenital primary developmental defects in 2 or more tissues originated from embryonic ectoderm. The skin and its appendages such as hair follicles, sweat gland, sebaceous gland and nail are the primarily involve tissues. Abnormalities in other tissues, e.g., ears, eyes, lips, mucous membranes of the mouth or nose, central nervous system are also associated with these diffuse and nonprogressive disorders. Ectodermal dysplasias are inherited diseases that comprise of a large heterogeneous group. To date, more than 192 distinct disorders have been described.[1]

Barber Say syndrome (BSS) is a rare ectodermal dysplasia (Prevalence <1/1 000 000) which presents at birth and is characterized by congenital generalized hypertrichosis, atrophic skin, ectropion and macrostomia.[2] Here we will describe a female neonate presented with signs in favor of BSS.

Case Report

One day old female neonate referred from periphery hospital due to facial and multiple organ anomalies. The patient was a product of normal vaginal delivery from 25 years old woman (G4 L2 A2) with gestational age 38 weeks and birth weight 2800 gr, without any significant problem during pregnancy and delivery. Two abortion in the past medical history of mother happened at about gestational age 3 months without any diagnosis. Four years old male sibling is alive and healthy with only history of mild hypertrichosis. Parents were 2nd degree relatives. There is no family history of any congenital anomaly.

On physical examination, the patient had normal vital signs with abnormal face (low hairline, coarse face, macrostomia, thin upper lip, bilateral severe ectropion and hypertelorism) [Figure 1], hypertrichosis particularly on the back [Figure 2] and lower extremities [Figure 3], senile skin appearance with mild redundant skin, particularly on the posterior of the neck, bilateral hypoplastic nipples and one area of mild skin atrophy (2 cm diameters) on the anterior mid-chest wall with irregular borders. Other physical exams were normal and there was no history of photosensitivity.

Small size secundum atrial septal defect (4 mm) in echocardiography was the other associated anomaly. The other para clinical evaluations, including brain and abdominal sonography, chest X-ray, complete blood count, venous blood gas, pulse oximetry, blood glucose, kidney function and electrolytes were normal.

At the time of admission, emergency ophthalmology consult was done with result, bilateral severe ectropion, erythematous lids, chemosis of conjunctiva, severe dry cornea, shallow anterior chamber, hypopigmented iris and normal fundoscopy. Eye care with artificial tear, eye lubricant

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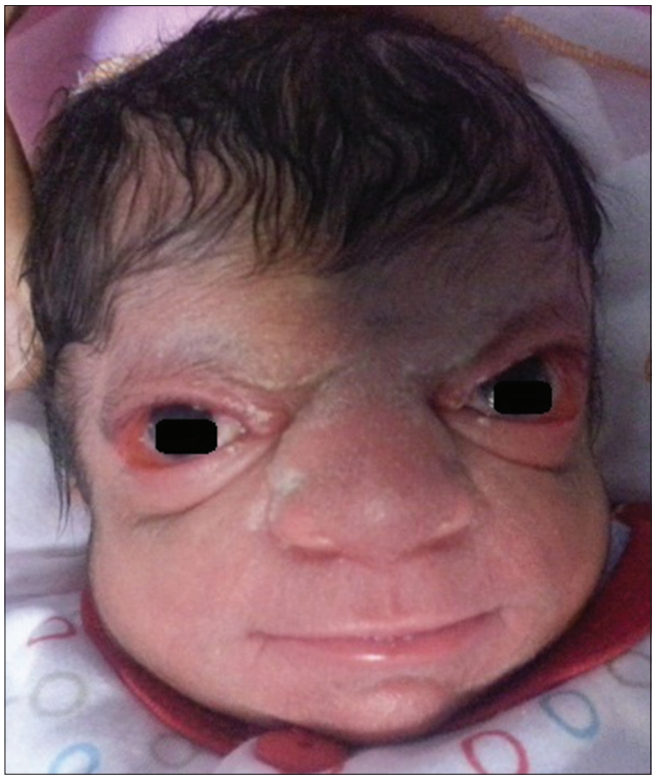

Figure 1: Low hair line, coarse face, macrostomia, thin upper lip, bilateral ectropion

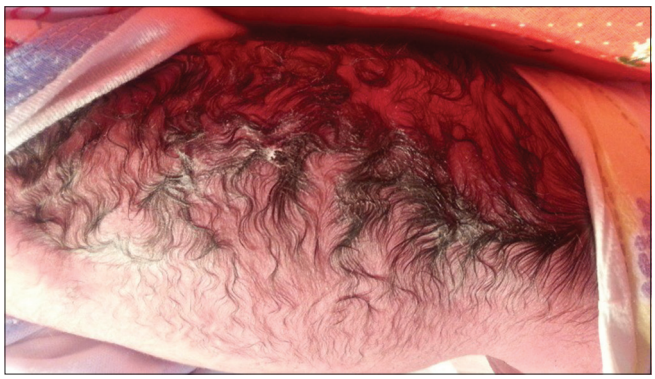

Figure 2: Hypertrichosis on back

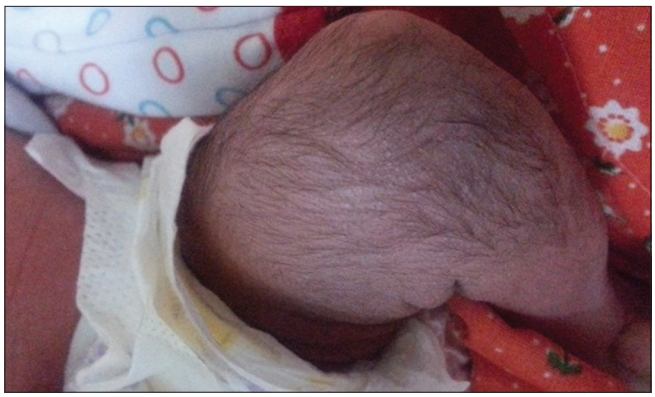

Figure 3: Hypertrichosis on lower extremity

and local antibiotics were started for the patient. Tarsoraphy was done on the second day of admission and she was discharged from hospital with breast feeding and good condition on the fourth day. After discharge, the patient's growth and development were normal and is under follow up of pediatrician, ophthalmologist and dermatologist for continuing her management.

Discussion

Congenital generalized hypertrichosis (CGH) is a heterogenous group of distinct conditions that differ in phenotype and genotype and has been seen as the major phenotype in 10 different genetic syndromes.[3] Our patient's characteristics are more compatible with BSS than other syndromes.

Barber Say syndrome has variable symptoms, consist of mild and severe presentations. The patient's characteristics are congenital generalized hypertrichosis, facial dysmorphism (typically with bilateral ectropion, absent or sparse eyebrows and lashes, hypertelorism/telecanthus, broad nasal bridge, bulbous nose, macrostomia, thin lips and misshapen ears), hyper laxity and the redundancy of the skin with deep folds, nipple hypoplasia and absence of mammary glands.[2]

The patient described in this case report presented with low hairline, coarse face, macrostomia, thin upper lip, bilateral ectropion and hypertelorism, hypertrichosis, senile skin appearance, hypoplastic nipples and one area of mild skin atrophy. The combination of these findings consistent with signs of BSS when compared with previous cases reported with this syndrome[4] [Table 1].

Parental consanguinity is reported in only two of the previous cases as in this present case. The interesting finding in our case was hypertrichosis in her brother who has no other sign of disease. Also secundum atrial septal defect was diagnosed in present case. Other findings in this case are the same as other case reports.

Autosomal dominant and autosomal recessive transmissions, as well as sporadic cases have been reported.[2] Dinulos suggests that at least some cases of Barber Say syndrome are caused by dominant mutations in the *TWIST2* gene.[9] The significance of the present case is that her brother has also mild hypertrichosis and no other congenital abnormality was reported in her family. So it is probable that this case is a sporadic one. Unfortunately, because the patient's parents did not permit gene analysis, we cannot have any suggestion in this field.

Acknowledgments

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Table 1: Comparison of present case with other previous reported cases

	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18
Physical findings at birth	Barber *et al.*, 1982[5]	David *et al.*, 1991[6]	Martinez - Santana *et al.*, 1993[7]	Mazzanti *et al.*, 1998[8]	Dinulos and Pagon (patient 1) 1999[9]	Cortés *et al.*, 2000[10]	Tenea and Jacyk 2006[11]	Haensel *et al.*, 2009[12]	Roche *et al.*, 2010[13]	Roche *et al.*, 2010[13]	Martins *et al.*, 2010[14]	Marchegiani *et al.*, 2015[15]	Marchegiani *et al.*, 2015[15]	Marchegiani *et al.*, 2015[15]	Marchegiani *et al.*, 2015[15]	Singh *et al.*, 2016[16]	Yohannan *et al.*, 2017[17]	Present case 2017
Sex	Female	Male	Female	Female	Male	Female	Male	Female	Male	Female		Male	Female	Female	Female	Female	Male	Female
Generalized hypertrichosis	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Ectropion	+	+	+	+	+	+	+	+	-	+	?	+	+	+	+	-	+	+
Telecanthus	+	+	+	+	+	+	?	+	-	+	+	+	+	+	-	-		+
Broad bulbous nose	+	+	+	+	+	+	+	-	+	+	-	+	+	+	+	+	+	+
Macrostomia	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Mother age	21	35	21	-	-	42				-							22	25
Parental consanguinity	-	-	+	-	-	-	-	-	-	-	-	-			+		-	Yes (2nd relative)
Familial recurrence	-	-	-	-	Yes (mother and son)	-			Yes (father and daughter)	Yes (father and daughter)	-					+	-	Mild hypertrichosis in sibling (male)
Birth weight (g)	2720	2550	2900	2200	-	3400	?	?	?	2650	?	?	?	?	?	?	3650	2800

+: Present, -: Not present, ?: No data on this finding

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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