CASE REPORT

A Case of Woolly Hair with Dextrocardia and Situs Inversus ("Faruk's Syndrome")

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ABSTRACT

Woolly hair (WH) and dextrocardia are a very rare condition along with situs inversus. A case of 18 years old young lady is found WH. No major abnormality is found. Her echocardiogram and electrocardiogram with the radiological finding of chest found normal except situs inversus. Hair bulb shows few dystrophic changes in WH due to mutation of the gene. Although WH patient's shows spares, fine and curly hair but with dextrocardia and situs inversus is the universal case that is why we call it "Faruk's Syndrome."

Key words: Dextrocardia, electrocardiogram, situs inversus, woolly hair

INTRODUCTION

A rare heart condition is dextrocardia, where heart is pointed toward the right side of chest instead of the left. In dextrocardia, people are not normal accordingly, which means abnormality is found in affected persons. People with dextrocardia usually do not suffer from medical emergency. They may be prone to bowel, esophageal, bronchial, and heart problems (as endocardial cushion defect and pulmonary stenosis). Few cardiovascular and pulmonary disorders associated with dextrocardia can be life threatening. Kartagener syndrome may be present with dextrocardia with situs inversus, and the patient may have sterility. Dextrocardia is usually found after 12,019 deliveries.[1] Autosomal-dominant woolly hair (WH) is a rare disorder where tightly curled hair is found. In WH of autosomal dominant, mutation of *KRT74* and *KRT71* gene mutations, and in autosomal recessive *LIPH* and *LPAR6* gene mutations found. Autosomal recessive WH can be syndromic may be accompanied by hyperkeratosis and cardiac abnormality.

CASE REPORT

Samira [Figure 1] 18 years unmarried young lady from Patuakhali district of Bangladesh is suffering from fine hair on her scalp since her childhood; her parents were not concerned about her hair at the 1st few years of her life. When she started her school life, her friends noticed that her hair is fine and density is lower than as usual. She took several types of medicine such as vitamin, anti-histamine, some ayurvedic medicine, and so many types of traditional medicine, but her condition was not improved. Then, she came to us with the complaint of the fine, thin, short, and low density of hair on her scalp. We took her family history, but no other members are suffering from the same type of problem. We did her Chest X-ray [Figure 2] where we found fundic gas shadow under the right hemidiaphragm; liver is on the left side apex of heart on the right side. Hence, we confirmed that it is a case of dextrocardia with situs inversus. Then, we did her electrocardiogram (ECG) [Figure 3] and ECG [Figure 4]. Both of the reports were fine, and no abnormality was detected except dextrocardia.

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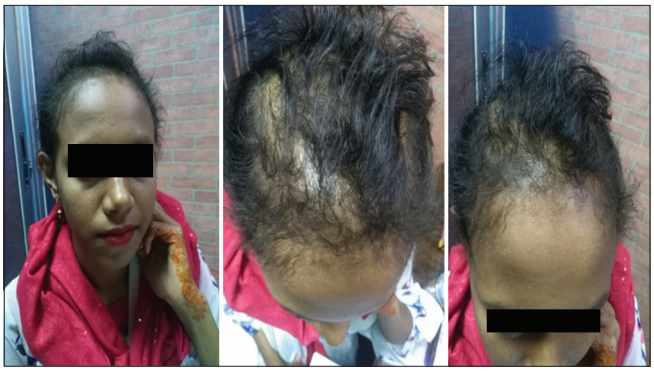

Figure 1: Fine, Silky hair of the patient

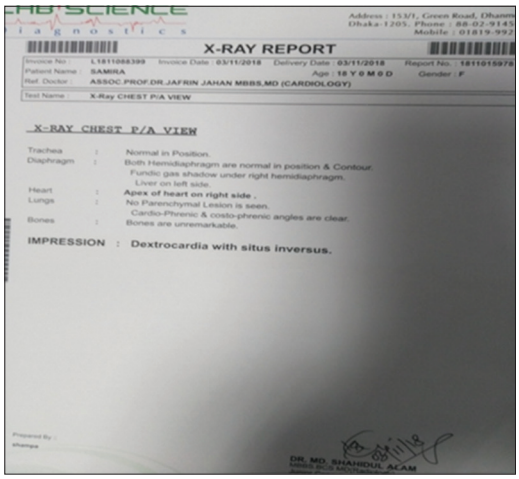

Figure 2: Chest X-ray report

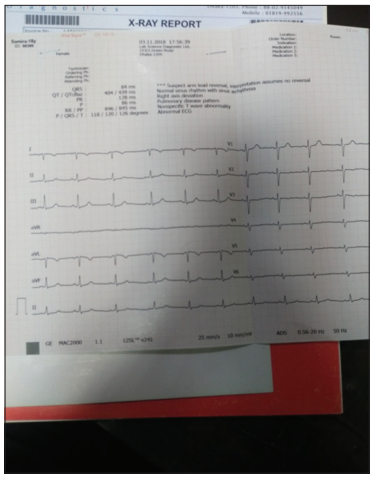

Figure 3: Electrocardiogram report

Figure 4: Echocardiogram report

DISCUSSION

WH is a rare congenital abnormality of the structure of the scalp hair characterized by tightly coiled hair involving part or the entire scalp occurring in an individual of non-Negroid origin. It was first observed and described by Gossage in 1907 in a European family.[1] Autosomal recessive WH/hypotrichosis (ARWH/H; OMIM 278150, 611452, 604379) is an inherited hair disorder characterized by sparse WHs on the scalp and occasionally sparse to absent eyebrows and eyelashes. Recently, causative genes for ARWH/H were identified, *LIPH* and *LPAR6*. The *LIPH* gene encodes phosphatidic acid (PA)-selective phospholipase A1α (PA-PLA1α) which hydrolyzes PA into 2-acyl lysophosphatidic acid (LPA) and free fatty acid. This LPA binds to a G protein coupled receptor LPA6, also known as P2Y5, encoded by the *LPAR6* gene, and the PA-PLA1α/LPA/LPA6 signaling pathway has been shown to play crucial roles in the hair follicle development [Table 1].[2,3]

Menkes disease is an X-linked lethal multisystem disorder caused by disturbances of copper distribution in different tissues due to mutation of p ATPase7 gene. The estimated prevalence of the disease is 1 in 100,000–1 in 250,000. The affected individual suffers from the malfunction of copper-containing enzymes resulting in multisystemic disturbances. Nervous system problems include gross mental retardation, convulsions, cortical atrophy, asymptomatic subdural effusion, gross truncal hypotonia, and progressive neurological deterioration, vascular problems with weak collagen tissues causes easy breakability; connective tissue abnormality gives rise to characteristics of steel, fuzzy, woolly, and sparse hair with easy pluckability.[4] Three cases of WH syndrome in the same family are reported. The abnormality is transmitted as a dominant autosomal trait. The main clinical features in our three patients were the fine, soft, and frizzy WH and generalized hypertrichosis.[5] WH is defined as an abnormal variant of tightly curled hair.[6] As compared with curly hair which is typically observed in African populations, WH does not grow well and stops growing at a few inches. The bulb portion of WH shows a dystrophic feature. In addition, the hair shaft is irregularly curved. Affected individuals with WH frequently show sparse scalp hair (hypotrichosis). There are both syndromic and non-syndromic forms of WH. The non-syndromic forms of WH can show either an autosomal dominant or recessive,

Table 1: Phenotypic variability in isolated hereditary hair loss disorders[3]

Disease	Genetic locus	Gene	Phenotype	HS abnormalities	HF abnormalities
Hypotrichosis simplex of scalp type 1 (HSS1/HYPT1)	18p11.22	*APCDD1*	Sparse, short and thin hair on the scalp and body. Eyebrows, eyelashes and beard hair are normal	No HS abnormality	NA
Hypotrichosis simplex of scalp type 2 (HSS2/HYPT2)	6p21.3	*CDSN*	Normal hair at birth, scalp hair growth retardation with diffuse hair loss, sparse hair on scalp, body. Eyebrows, eyelashes, and beard hair are normal	No HS abnormality	NA
Dominant hereditary hypotrichosis 3 and WHs (HYPT3)	12q13	*KRT74*	Sparse scalp hair. Coarse, lusterless, tightly curled WH. Eyebrows, eyelashes, and beard hair are normal	Dystrophic anagen hair. HS twisted with tapered distal ends	NA
Marie Unna hereditary hypotrichosis 1/ hypotrichosis 4 (MUHH1/HYPT4)	8p21.2	*U2HR*	Slow growing sparse and fragile hair	NA	NA
Marie Unna Hereditary hypotrichosis 2/ hypotrichosis 5 (MUHH1/HYPT5)	1p13.3	*EPS8L3*	Sparse to absent scalp hair at birth. Wiry, irregular hair in childhood. Thin eyebrows, eyelashes	NA	Markedly reduced mature HFs
Localized autosomal recessive hypotrichosis 1/hypotrichosis 6 (LAH1/HYPT6)	18q12	*DSG4*	Sparse scalp hairs, absence of eyebrows and eyelashes, normal axillary and pubic hairs, follicular papules on scalp	Coarse, short, brittle and fragile HS, trichoschisis, pili torti, uneven HS diameter, tapered end	NA
Localized autosomal recessive hypotrichosis 2/hypotrichosis 7 (LAH2/HYPT7)	3q27.2	*LIPH*	Sparse, thin, fragile, and short scalp hair. Woolly, tightly curled, light-colored scalp hair. Normal to sparse eyebrows, eyelashes, axillary, and body hair	Twisted HS with tapered distal ends	Comedo-like HF with hyperkeratinization
Localized autosomal recessive hypotrichosis 3/hypotrichosis 8 (LAH3/HYPT8)	13q14	*LPAR6*	Tightly curled, woolly, fragile and slow-growing hair, normal to sparse eyebrows and eyelashes, popular lesions on the occipital region	Absent root sheath component in the bulb region	Small-sized HF with dilation of lower infundibula, comedo-like HF with hyperkeratinized infundibula
Localized autosomal recessive hypotrichosis 4/hypotrichosis 9 (LAH4/HYPT9)	10q11.23-q22.3	Yet to be discover	Sparse, thin and slightly brown hair on scalp, arms, and legs. Normal eyebrows and eyelashes	NA	NA
Localized autosomal recessive hypotrichosis 5/hypotrichosis 10 (LAH5/HYPT10)	7p22.3-p21.3	Yet to be discover	Complete absence of hair at birth. Eyebrows, eyelashes, and body hairs are sparse. Papules on scalp	NA	Markedly reduce the number of HF

(Contd...)

Table 1: *(Continued)*

Disease	Genetic locus	Gene	Phenotype	HS abnormalities	HF abnormalities
Hypotrichosis simplex of scalp type 3 (HSS3/HYPT11)	1q32.1	*SNRPE*	Thin scalp and axillary hair to complete absence of scalp and body hair, sparse to absent eyebrows, normal pubic hair	No abnormalities of HS	Vellus type of HF
Hypotrichosis simplex of scalp type 4 (HSS4/HYPT12)	13q12.2	*RPL21*	Normal hair at birth, hair loss began at 2–6 months of age, absent to sparse scalp hair, eyebrows, eyelashes, body hair, and axillary and pubic hairs	No HS anomaly	HF significantly decreased in number and size
Hypotrichosis with recurrent skin vesicles	18q12.1	*DSC3*	Sparse scalp hair, absence of eyebrows, eyelashes, axillary and body hair, skin vesicles of <1 cm in diameter on scalp and skin	NA	Slight follicular plugging
Digenic autosomal recessive hypotrichosis	16q22.1 and 12q21.2-q22	*CDH3* and an unknown gene	Absent or sparse scalp hair, normal eyebrows, eyelashes, axillary and body hair	NA	NA

inheritance pattern.[7] Contrary to other entities within the WH group, diffuse partial WH so far has not been associated with any other disorder, particularly palmoplantar keratosis and cardiac anomalies.[8] Eight cases of situs inversus totalis were detected in the Atomic Bomb Casualty Commission-Japanese National Institute of Health Adult Health Study sample of Hiroshima and Nagasaki, Japan. The prevalence of situs inversus totalis was 1/4100 population.[9] Further observations and studies into the molecular and genetic basis of dextrocardia, situs inversus with WH (Faruk's Syndrome is a condition which is combined with dextrocardia, situs inversus, and WH) are required to confirm or exclude the possible connection.

CONCLUSION

WH is an autosomal disease. It is a unique case that we could call it "Faruk's Syndrome." Much more study is needed for further evaluation.

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