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Case presentation

Generalized woolly hair with diventricular arrythmogenic cardiomyopathy: a rare variant of Naxos disease

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Abstract

Woolly hair may occur as an isolated problem of cosmetic concern or can be a part of a systemic disease (woolly hair syndrome) with underlying fatal cardiomyopathy. Two characteristic associations of woolly hair syndrome are Naxos disease and Carvajal syndrome. Naxos disease is characterized by woolly hair, palmoplantar keratoderma, and arrythmogenic right ventricular cardiomyopathy.

In this report we describe a case of a young girl who presented with heart failure and was subsequently diagnosed as a case of generalized woolly hair with biventricular arrythmogenic cardiomyopathy.

Our case represented a rare variant of Naxos disease in the advanced stage of arrythmogenic right ventricular cardiomyopathy; biventricular failure may occur with involvement of the interventricular septum and left ventricle causing congestive heart failure.

Keywords: woolly hair, Naxos disease, arrythmogenic cardiomyopathy

Introduction

The term woolly hair denotes an abnormal structural defect of scalp hair that is fine, with tightly coiled curls [1, 2]. Three forms of woolly hair do exist: autosomal dominant (hereditary form), autosomal recessive (familial wooly hair), and localized nonhereditary woolly hair in the form of woolly hair nevus [1, 3]. Woolly hair may occur as an isolated problem of cosmetic concern or can be a part of a systemic disease (woolly hair syndrome) with underlying fatal cardiomyopathy [1, 3]. In this report we describe a case of a young girl who presented with heart failure and subsequently was diagnosed as a case of generalized woolly hair with biventricular arrythmogenic cardiomyopathy.

Case synopsis

A four-year-old girl presented to us with a history of acute onset of exertional breathlessness for the preceding 5 days. Her past medical and birth history was uneventful. On examination the girl was orthopneic; her pulse rate was 110/ min, of low volume, and irregular in nature. Her blood pressure was 90/60 mm of mercury. There was dependant pedal edema, engorged pulsatile neck vein, tender soft hepatomegaly, and bibasilar pulmonary rales with S3 (third heart sound) gallop rhythm. It was noteworthy that she had curly easily pluckable hair, lighter in color (Figure 1).

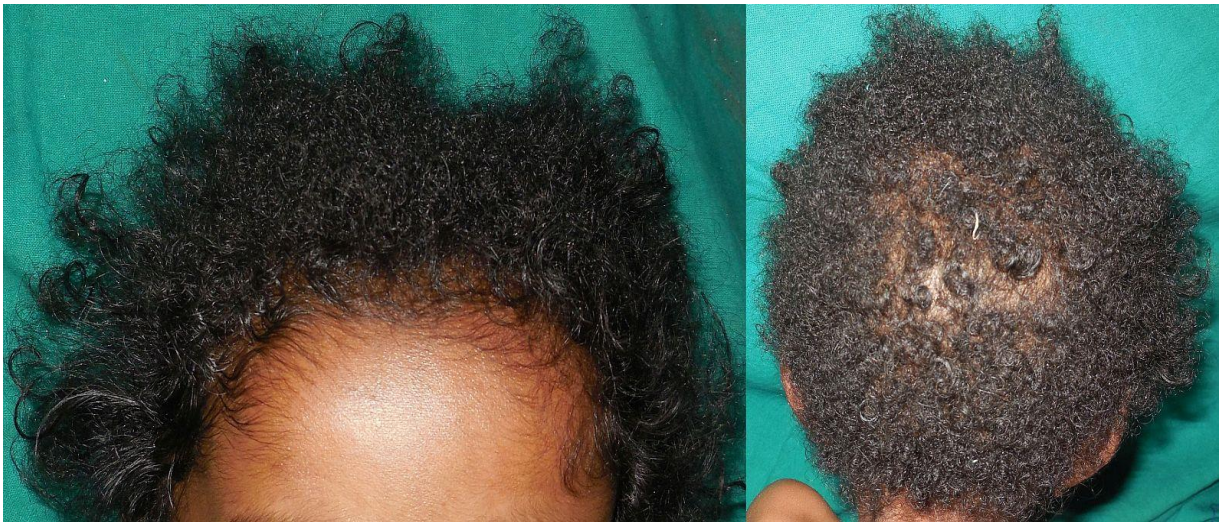

Figure 1. Curly hair (lighter in color) on the scalp.

However, her eyebrows, nails, and teeth were normal. Her palms were xerotic and thickened and the soles were dry, thickened, and fissured. We also noticed a plantar clavus (Figure 2).

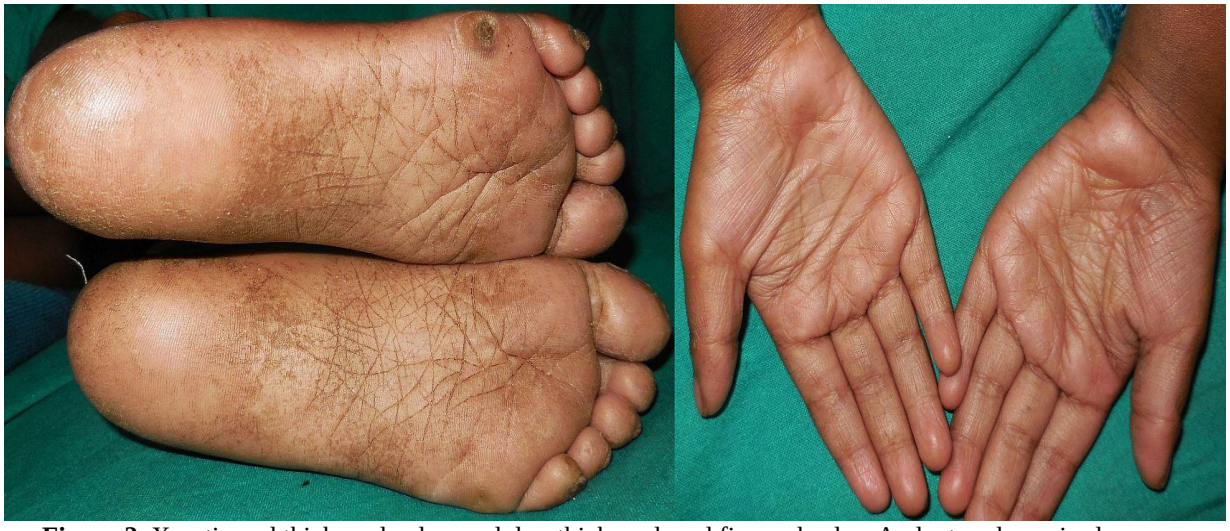

Figure 2. Xerotic and thickened palms and dry, thickened, and fissured soles. A plantar clavus is also seen.

Further query revealed that she had a history of consanguineous parentage and several family members over generations reportedly had a similar type of hair. It is worth mentioning that one of her brothers died at the age of six years owing to sudden cardiac death and our patient had suffered several episodes of syncope.

Chest X-ray PA view showed an enlarged cardiac silhouette. Frequent ventricular ectopic beats, biatrial enlargement, and intraventricular conduction defects were noted in different leads of the electrocardiography (ECG) (Figure 3).

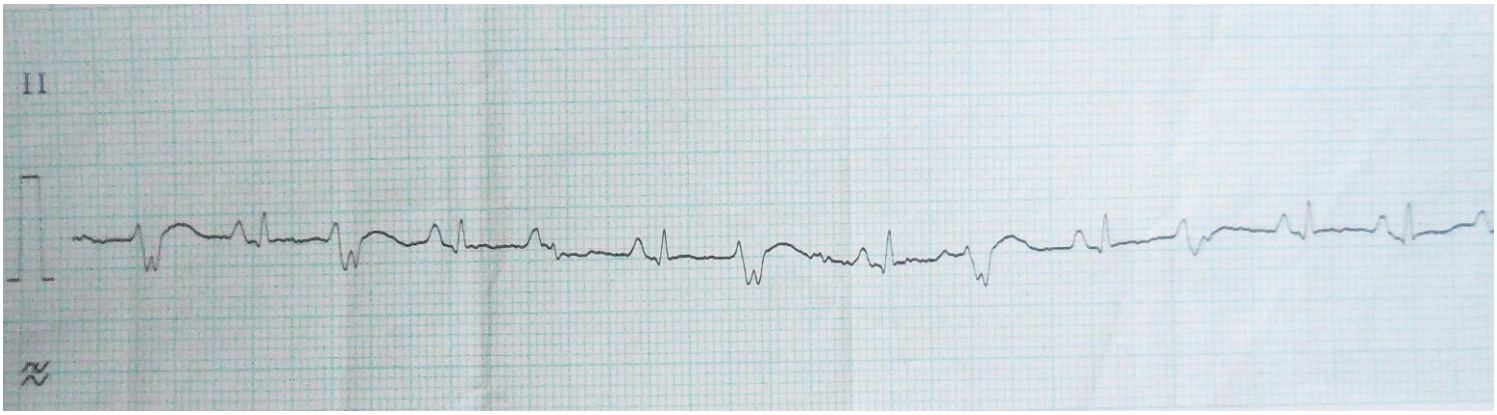

Figure 3. Electrocardiography (long lead II) showing frequent ventricular ectopic beats.

Transthoracic echocardiography revealed dilatation of all of the four chambers of the heart with global hypokinesia and biventricular systolic dysfunction. In addition, moderate mitral and tricuspid regurgitation was present. Her ejection fraction was 48.20% (Figure 4.)

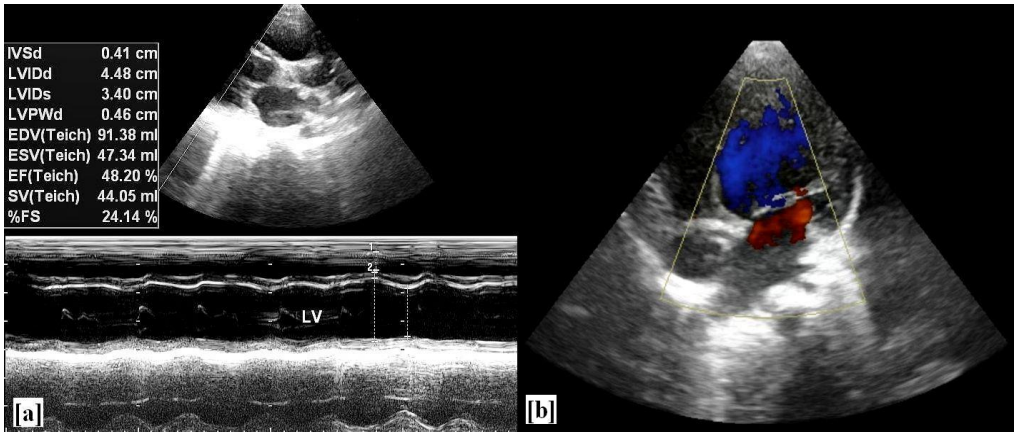

Figure 4. (a) Parasternal long axis view M-mode study showing dilated LV with reduced EF (b) Apical 5-chamber view with color flow doppler showing dilatation of all cardiac chambers.

Computed tomographic scan of the brain and electroencephalogram were normal. Complete hemogram and biochemical panels were noncontributory. Light microscopic examination of scalp hair showed the presence of tight spirals and reduction of the diameter in comparison to the normal hair (Figure 5).

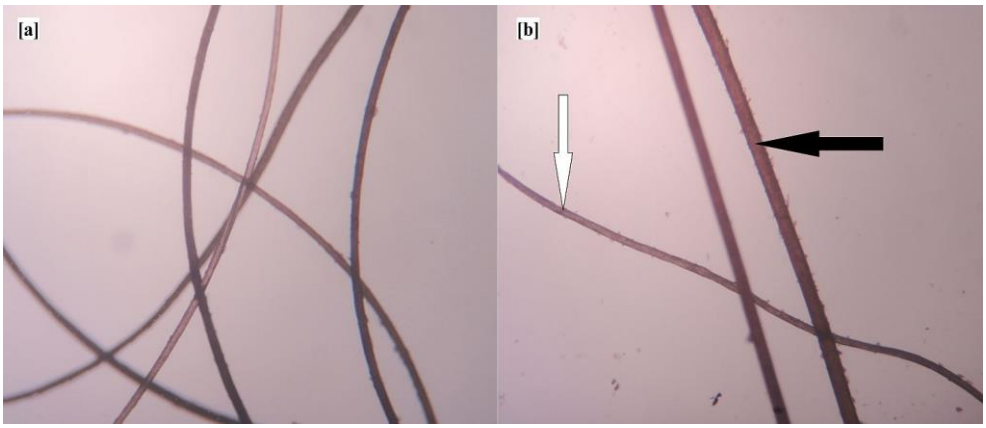

Figure 5. Light microscopic examination of scalp hair showing (a) the presence of tight spirals and (b) reduction of the diameter of the woolly hair (white arrow) in comparison to the normal hair (black arrow).

The thickness of the hair was assessed by hair analysis software (Hairsys, South Korea). The hair of the patient had an average thickness of 0.00497 mm (normal hair thickness: 0.05-.15 mm).

Based on characteristic features, a diagnosis of woolly hair syndrome with biventricular arrythmogenic cardiomyopathy was made. She was treated conservatively in the intensive care unit and recovered from cardiac failure within a couple of weeks.

Discussion

Woolly hair is extremely curly, with reduced average diameter of hair. Compared to normal curly hair that is observed in some populations, woolly hair grows slowly and stops growing after a few inches [3]. Light microscopy of hair in all the woolly hair disorders reveals non-specific features, structural anomalies, trichorrhexis nodosa, and tapered ends [1, 3].

Isolated woolly hair is inherited in an autosomal dominant fashion, which may be associated with enamel hypoplasia, ocular defect, deafness, Noonan syndrome, icthyosis, and keratosis pilaris atropicans [2].

Two characteristic associations of woolly hair syndrome are Naxos disease and Carvajal syndrome [4]. Naxos disease is an autosomal recessive disorder owing to mutation in the plakoglobin gene, gene map locus 17 q21 [2]. Naxos disease is characterized by woolly hair, palmoplantar keratoderma, and arrythmogenic right ventricular cardiomyopathy (ARVC) [4]. Woolly hair appears from birth, whereas palmoplantar keratoderma develops during infancy when infants start to use their hands and feet. The cardiomyopathy clinically manifests by adolescence and shows 100% penetrance.

On the other hand, Carjaval syndrome usually presents at a younger age, with predominantly left ventricular involvement, early morbidity, and clinical overlapping with dilated cardiomyopathy [4]. It occurs owing to mutation in the *desmoplakin* gene, map locus 6p24 [2]. We could not perform a genetic study of our patient because of local unavailability, which was a limitation of our report. However a pedigree chart of our patient pointed towards an autosomal recessive mode of inheritance. Presence of woolly hair in multiple family members over several generations and a history of sudden cardiac death in a family member were also notable features in the present case.

Our patient had biventricular arrythmogenic cardiomyopathy, which was not typical of Naxos or Carjaval syndrome. However, about 27% cases of Naxos disease may have additional left ventricular involvement [5]. Baykan et al previously reported three such cases [4].

Our patient represented a variant of Naxos disease; the advanced stage of ARVC biventricular failure might occur with involvement of the interventricular septum and left ventricular failure causing congestive heart failure [6].

Conclusion

In this report we seek to emphasize the importance of detailed cardiovascular examination and examination of the other family members in every patient with woolly hair syndrome, considering its association with cardiomyopathy. When an early diagnosis is made, the life expectancy of such cases may be increased by appropriate treatment of heart failure and arrhythmias.

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