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CASE REPORT

Hypertrichosis cubiti, a case report and literature review

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Key Clinical Message

Hypertrichosis cubiti is an uncommon congenital hypertrichosis with links to genetic syndromes, both autosomal dominant and recessive, with variable penetrance and expressivity. It may also present in sporadic cases with no phenotypic abnormalities or family history.

Keywords

Elbow, hypertrichosis.

Introduction

Hypertrichosis cubiti, also known as hairy elbows syndrome, is an uncommon type of congenital hypertrichosis with long vellus hair in the elbow area. There are only 50 patients reported in the literature since 1970, when Beighton reported his first cases [1]. The mode of inheritance remains unclear, with reports suggesting either an autosomal recessive, or autosomal dominant form with variable penetrance and expression, or a spontaneous mutation. Sporadic cases with no reported abnormalities have also been reported.

Case Report

A 4-year-old child was seen with her parents about excessively hairy elbows, first noticed around age three and a half. She was an only child, with no family history of hypertrichosis, was otherwise well, on no medications and had no known allergies. Pregnancy and delivery history were normal. Her immunizations were up to date and she was otherwise developmentally normal. Other than the hairy elbows, her parents have no other concerns about her health.

On examination, she was a well child, with no unusual facial or dysmorphic features. Her height was 108 cm (90th centile) and weight was 19.5 kg (90th centile). There was fine vellus-type blonde hair over her arms, legs, and back. The longest hair in the elbow area measured up to 5 cm and affected the lower third of the arm and the upper third of the forearm (see Figs. 1 and 2). There was also fine vellus-type blonde hair on her knees which were uniform in length of about 1.5 cm. The fine vellus-type hair on her lumbosacral spine centrally and neck were of a uniform length of about 1.5 cm.

There were macular erythematous patches on her glabellar area, occipital scalp, and centrally over the cervical spine, which were thought to be capillary malformations. However, due to multiple vascular marks, there was a concern about possible underlying malformations such as a faun tail or intracranial hemangiomas.

Her full blood count, urea, electrolytes and creatinine, liver function tests, thyroid-stimulating hormone, iron studies, antinuclear antibodies (ANAs), hormone profiles, and serum electrophoresis were all normal (see Table 1 for blood test results). An X-ray showed a bone age of 3 years 6 months when her chronological age is 4 years 3 months. A magnetic resonance imaging (MRI) study of her spine was performed which excluded any spinal or intracranial abnormalities associated with her vascular birthmarks.

Discussion

Hypertrichosis cubiti was first described by Beighton in 1970 [1]. To our knowledge there are 50 documented

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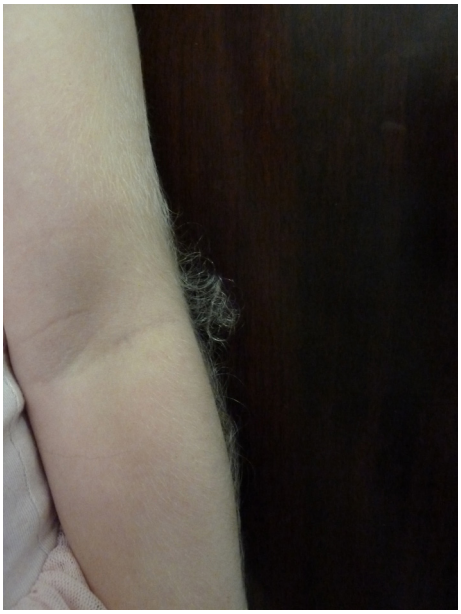

Figure 1. Left elbow.

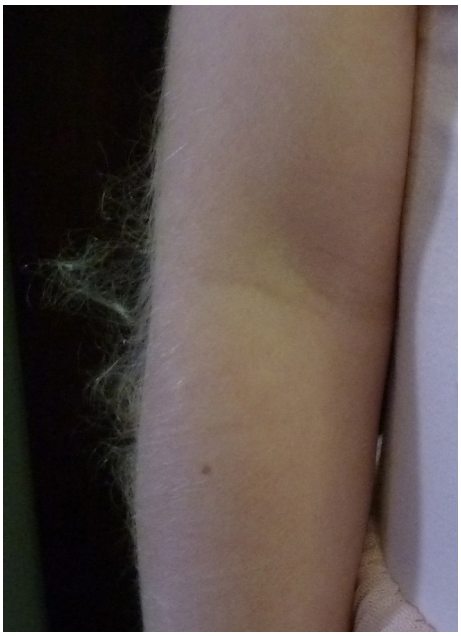

Figure 2. Right elbow.

Table 1. Blood test results.

Test	Results	Normal range/reference interval
Hemoglobin	128 g/L	115–150
White cell count	6.8 × 10^9/L	4.0–12.5
Platelets	264 × 10^9/L	150–480
Urea	5.5 mmol/L	1.8–6.0
Creatinine	40 μmol/L	15–50
Thyroid-stimulating hormone	1.8 mIU/L	0.5–4.5
Cortisol	127 nmol/L	200–600[1] (Blood collected 10 AM)
Free Androgen Index	<0.1	Females nonpregnant: 20–120[2]
Sex Hormone-Binding Globulin	193.4 nmol/L	19.0–120.0[2]
Dehydroepiandrosterone sulfate DHEAS	<0.1 μmol/L	0.0–0.5
Testosterone	<0.4 nmol/L	0.0–1.0
Estradiol	<18 pmol/L	<18–80
Luteinizing hormone	<0.1 IU/L	0.0–4.4
Follicle-stimulating hormone	1.3 IU/L	0.2–7.5
Prolactin	191 mIU/L	0–760
Adrenocorticotropic hormone	1.5 pmol/L	<11
Growth hormone	2.4 mIU/L	<13
Insulin	4 mU/L	<10
Intact parathyroid hormone	1.9 pmol/L	1.5–7.6
IGF-1 (Somatomedin C)	13 nmol/L	3–17

[1]Diurnal variation: morning 200–600 nmol/L; afternoon approximately one-third of morning value.
[2]Expected results as Free Androgen Index and Sex Hormone-Binding Globulin are not well characterized in this age range.

cases as of 2014 (Table 2). Several cases are mentioned in other languages (French [2], Spanish [3]), or are purely observational [4, 5]. Only one documented biopsy result has been published [6] indicating normal hair follicles with an increased percentage of hairs in anagen phase (90%).

Different inheritance patterns have been postulated, including a familial pattern with either an autosomal dominant or autosomal recessive inheritance, with variable penetrance and expressivity [1, 7–12]. Other theories include primary nevoid hypertrichosis [6, 13] or somatic hypertrichosis mosaicism [14, 15]. Some links to syndromes such as the Weill–Marchesani syndrome [1], Wiedemann–Steiner Syndrome [16], or Floating-Harbor syndrome [9] have been suggested, but are inconclusive. A number of reports link hypertrichosis cubiti to short stature and/or developmental delay [1, 2, 8, 9, 11, 12, 15–19], but this so far has been reported only in cases which were thought to have a possible link to a syndrome [1, 9, 16, 20]. In the sporadic cases, endocrine and chromosomal studies have been normal [13, 21–24] and are not linked to mental or physical abnormalities. The excess hair often resolves by adolescence [7, 8, 13, 21, 22], except for one case that persisted into adulthood [10]. It has also been suggested that this condition is far more prevalent but under-reported, for instance in male chil-

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Table 2. Case reports of hypertrichosis cubiti

Authors	Year	No of patients	Proposed inheritance	Age years	Family history	Short stature	Associated anomalies
Andreev, Stransky	1979	1	Nevoid condition, inheritance unclear	5	No	No	No
Beighton	1970	2	Autosomal recessive or autosomal dominant with variable expression (Weill–Marchesani Syndrome)	12, 13	Yes, Father and grandfather	Yes	Faun tail, regressed, short fingernails
Cambiaghi, Pistretto, et al.	1998	4	Autosomal dominant with variable penetrance and expression	4 to 9 years	Yes, Father and grandmother	No	No
Coleman	1994	1	Inheritance unclear	5	No	No	No
Di Lernia, Neri, et al.	1996	5	Autosomal dominant with variable penetrance and expression	7, 10 plus 3 adults	Familial	Yes	No
Edwards, Crawford, et al.	1994	1	Somatic mosaicism	3	No	No	Asymmetry of face, developmental delay
Escalonilla, Aguilar, et al.	1996	1	Variable inheritance pattern or sporadic	8	No	No	No
Fernandez-Crehuet P, Ruiz-Villaverde, Serrano	2013	1	Sporadic, nevoid hypertrichosis	6	No	No	No
Flannery, Fink et al.	1989	1	Genetic, unclear	12, 13	Amish ancestors	Yes	Facial anomalies, Hypotonia, Developmental delay
Jones, Dafou, et al.	2012	6	De novo mutations of MLL (Wiedemann–Steiner Syndrome)	N/A	De novo mutations	Yes 5 of 6	Facial anomalies, intellectual disability.
Koc, Karaer, et al.	2007	1	Autosomal recessive (Allelic variant of Floating-Harbor syndrome)	8	Consanguinous parents	Yes	Facial anomalies, microcephaly, joint hyperlaxity, developmental delay
Leon-Muinos, Montegudo, et al.	2009	1	No comment (Spanish)	5		No	
Lestringent, Frossard	1997	1	Autosomal recessive OR neomutation, autosomal dominant	28	Yes, mother	No	No
MacDermott, Patton, et al.	1989	4	Genetic heterogeneity in transmission. 2 cases autosomal dominant, 2 cases autosomal recessive	12.5, 7, 8, adult	2 sporadic, 2 familial	Yes	Facial anomalies, skeletal abnormalities
Martinez de Lagran, Gonzalez-Perez, et al.	2010	1	Unclear, familial or sporadic	5	No	No	
Miller, Matthew, Yeager, Josef	1995	1	Sporadic	7	No	No	
Nardello, Mangano, et al.	2008	1	No comment			Yes	Infantile spasms, behavior disorders, and cerebral hemisphere asymmetry
Plantin, Le Roux, et al.	1993	1	No comment (French)	10	Sporadic	Yes	Intrauterine growth retardation
Polizzi, Pavone, Ciano, et al.	2005	3	Somatic mosaicism due to postzygotic mutation or paradominant inheritance or revertant mosaicism	7, 7, 11	Sporadic	No; Yes; Yes	No; Facial anomalies and developmental delay; Dysmorphic

(*Continued*)

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Table 2. Continued.

Authors	Year	No of patients	Proposed inheritance	Age years	Family history	Short stature	Associated anomalies
							features, developmental delay.
Rosina, Pugliarello, et al.	2006	1	Unclear, genetic	8	Sporadic	Yes	No
Rudolph	1985	1	No comment			No	
Schwarze, Loche, et al.	1999	4	Unclear	10	No	No	No
Vashi, Mancini, Paller	2001	2	Unknown origin	3.5, 6	No	No	No
Visser Beemer, Veenhoven, De Nef	2002	2	Either autosomal recessive or autosomal dominant	N/A	No	Yes	Facial anomalies, intellectual disability.
Warner	1980	1	Observation, no comment	7	Unclear	No	No
Yuste-Chave, Zafra-Cobo, et al.	2007	2	May be part of a complex syndrome with varying manifestations or sporadic	6, 10	Sporadic	No	No
		50					

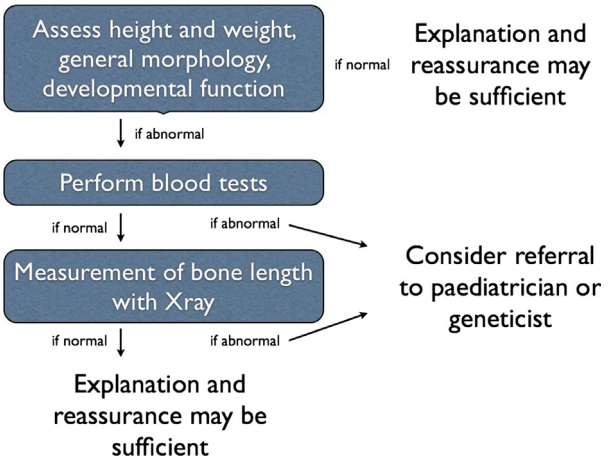

Figure 3. Management flow chart.

dren of dark-haired races, and in some of these cases it may be considered part of the range of physiological difference, instead of a pathological problem [22, 25]. These are summarized in Table 2.

Some cases of hypertrichosis cubiti could be linked to an undefined genetic syndrome, but sporadic cases without any other abnormalities may represent a cosmetic problem rather than something more sinister. Avoidance of further tests is encouraged if the remainder of the history and examination is normal (see Fig. 3). Reassurance of parents and advice regarding hair removal or bleaching would be appropriate for children with sporadic hypertrichosis cubiti. Hair removal options should be discussed with care to minimize discomfort and cost [26]. Spontaneous resolution of nonsyndromic cases of hypertrichosis cubiti tend to occur by adolescence [1, 3, 6–8, 13, 19, 21, 22, 25] and follow-up to check resolution may be appropriate.

Conflict of Interest

None declared.

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