**Hypertrichosis cubiti, a case report and literature review**

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**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
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-

**Table 1. Blood test results.**

| Test                                    | Results  | Normal range/reference interval |
|-----------------------------------------|----------|---------------------------------|
| Hemoglobin                              | 128 g/L  | 115–150                         |
| White cell count                        | 6.8 x 10^9/L  | 4.0–12.5                       |
| Platelets                               | 264 x 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 (Blood collected 10 AM) |
| Free Androgen Index                     | <0.1     | Females nonpregnant: 20–120     |
| Sex Hormone-Binding Globulin            | 193.4 nmol/L | 19.0–120.0                     |
| 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 mIU/L  | <10                             |
| Intact parathyroid hormone              | 1.9 pmol/L | 1.5–7.6                        |
| IGF-1 (Somatomedin C)                   | 13 nmol/L | 3–17                            |

1Diurnal variation: morning 200–600 nmol/L; afternoon approximately one-third of morning value.
2Expected results as Free Androgen Index and Sex Hormone-Binding Globulin are not well characterized in this age range.
### 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 | Faun tail, regressed, short fingernails |
| Di Lernia, Neri, et al. | 1996 | 5 | Autosomal dominant with variable penetrance and expression | 7, 10 plus 3 adults | Familial | Yes | No |
| Coleman | 1994 | 1 | Inheritance unclear | 3 | No | Asymmetry of face, developmental delay |
| Edwards, Crawford, et al. | 1994 | 1 | Somatic mosaicism | 12, 13 | Amish ancestors | Yes | Facial anomalies, Hypotonia, 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 | De novo mutations | No | No |
| 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, Hypotonia, Developmental delay |
| 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-Munos, Montegudo, et al. | 2009 | 1 | No comment (Spanish) | 5 | No | No | 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 | No; Facial anomalies, skeletal abnormalities |
| Martinez de Lagran, Gonzalez-Perez, et al. | 2010 | 1 | Unclear, familial or sporadic | 5 | No | No | No |
| Miller, Matthew, Yeager, Josef | 1995 | 1 | Sporadic | 7 | No | No | No |
| Nardello, Mangano, et al. | 2008 | 1 | No comment | 10 | Sporadic | Yes | Intrauterine growth retardation |
| Plantin, Le Roux, et al. | 1993 | 1 | No comment (French) | 10 | Sporadic | Yes | No; Facial anomalies and developmental delay, Dysmorphic |
| Polizzi, Pavone, Ciano, et al. | 2005 | 3 | Somatic mosaicism due to postzygotic mutation or paradoiminant inheritance or revertant mosaicism | 7, 7, 11 | Sporadic | No; Yes | Yes |

(Continued)
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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