Do you know this syndrome?*

Você conhece esta síndrome?

Laura Maria Andrade Silveira[1]
Isadora Rosado do Amaral[1]
Andréia Nogueira Ramos[2]
Vitoria Regina Pedreira de Almeida Rêgo[3]

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CASE REPORT

The case described here refers to a female patient, black, 2-years-old, accompanied by her mother who reported the increase of hair in the external ear and face of her baby since birth. At 5 months, the thin, light blond lanugo hair covered the entire face, armpits, groin and lower limbs. The parent denied the presence of lanugo hair on the mucous membranes, palms and plants. The patient was born by cesarean delivery at term and had had normal psychomotor development, but with delayed dentition. The dental units available were deformed. The child's parents were second cousins. The mother denied the use of drugs or alcohol during pregnancy, as well as any family members with similar signs or symptoms. On examination, the patient had long thin and light-colored hair on the face, especially on the upper lip and mandibular regions, axillae, external genitalia, back and lower limbs (Figures 1 and 2). The presence of five abnormal dental units was observed. The patient, showing no other associated anomalies, was diagnosed with Congenital Hypertrichosis Lanuginosa, and referred for evaluation to the dentistry, genetics and ophthalmology clinics.

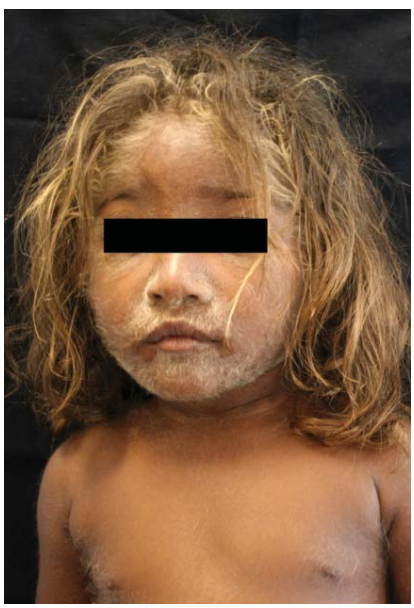

FIGURE 1: Child with lanugo hair distributed over entire body, sparing mucosal, palms and soles

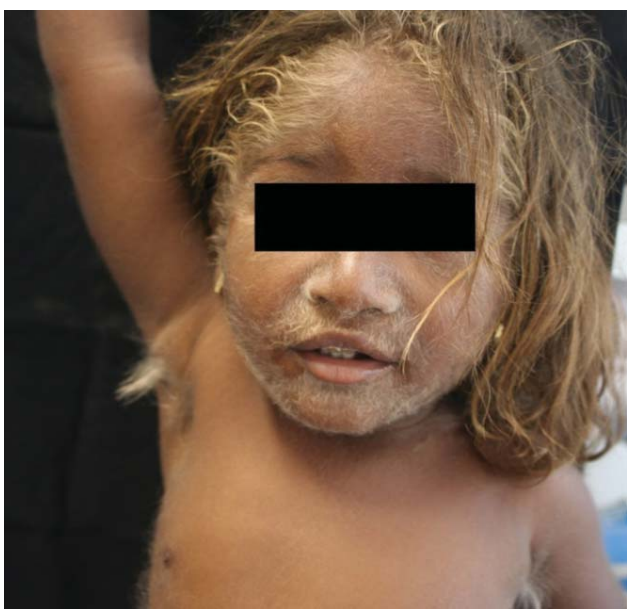

FIGURE 2: Detail of long lanugo hair in the axilla

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[1] MD - Resident in Dermatology- Federal University of Bahia. Prof. Edgar Santos Hospital Complex – Salvador (BA), Brazil.
[2] MD – Dermatologist - Private Clinic – São Paulo (SP), Brazil.
[3] Master- Chief of Dermatology Center, Prof. Edgar Santos Hospital Complex. Federal University of Bahia (COMHUPES-UFBA), Professor at Federal University of Bahia (UFBA) – Salvador (BA), Brazil.

DISCUSSION

Congenital Hypertrichosis Lanuginosa (CHL) is a rare autosomal dominant disorder with variable expressivity, characterized by excessive lanugo hair, sparing only mucous membranes, palms and soles.[1] It may be associated with other congenital abnormalities, mainly dental, but can also affect the ears, as well as being a symptom of glaucoma, pyloric stenosis, skeletal disorders and, more rarely, mental retardation.[2] CHL is a sporadic disease.[3] Clinical manifestations occur because there is no replacement of the lanugo hair by vellus or terminal pili.[4] Some 50 cases have been reported in the literature, with an estimated incidence of 1:10,000,000. The condition may get worse during childhood and puberty, causing serious social and aesthetic impacts. Aesthetic correction of the excessive hair is the priority treatment, although the results are disappointing. Several depilatory procedures can be used such as hair removal lasers, creams, razors etc, to improve the patient´s appearance. Published reports exist of the use of laser Q-switched Nd: YAG, effecting a reduction of 40-80% of the hair density, with little pain and low fluences.[5] Management of CHL also includes dental care, while genetic counseling is mandatory.

The differential diagnosis embraces diseases related to hypertrichosis in childhood including: universal hypertrichosis, Ambras syndrome, hypertrichosis linked X (Table 1).[6]

Recognition of the disease is important for screening other potential abnormalities and instituting genetic counseling. ❑

Table 1: Differencial diagnosis of hypertricosis in childhood

Name	Genetic Background	Clinical Features
Congenital Hypertrichosis Lanuginosa	Autosomal dominant	Fine, gray to blond lanugo hair.
Universal hypertrichosis	Autosomal dominant	Thicker, long hair most prominent on face, back and proximal extremities. Increases during childhood and tends to persist.
Ambras syndrome	Autosomal dominant (8q22)	Fine, silky long hair uniformly distributed on the face, ears and shoulders Persists for life Minor facial abnormalities, dental and supernumerary nipples
X-linked hypertrichosis	X-linked (Xq24–q27.1)	Curly, short, dark hair on the face and upper body Anteverted nostrils, prognathism,dental anomalies, deafness

Adaptated: Bolognia JL, et al. 20106

Abstract Congenital Hypertrichosis Lanugionsa is a rare autosomal dominant genetic disorder, with fewer than 50 cases reported in the literature. It is characterized by excessive lanugo hair, sparing only the mucous membranes, palms and soles. It may be associated with other organic abnormalities and should form part of the dermatologist´s current knowledge. We discuss some aspects of the syndrome in question arising from the case report of a 2-year-old female patient, black, with classic clinical presentation, with no other associated congenital abnormalities.
Keywords: Congenital abnormalities; Hypertrichosis; Laser

Resumo A hipertricose Lanugionsa Congênita é uma desordem genética rara, autossômica dominante, com menos de 50 casos descritos na literatura. É caracterizada por pêlo lanugo excessivo, poupando apenas membranas mucosas, palmas e plantas. Pode estar associada a outras anormalidades orgânicas, devendo ser de conhecimento do dermatologista. Discutiremos aspectos da síndrome em questão a partir do relato de caso de uma paciente do sexo feminino, negra, 02 anos, com apresentação clínica clássica, sem outras anormalidades congênitas associadas.
Palavras-chave: Anormalidades congênitas; Hipertricose; Laser

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MAILING ADDRESS:
Laura Maria Andrade Silveira
Ambulatório Magalhães Neto
Rua Padre Feijó, 240/3o Andar.
Canela
40110-170 Salvador, BA.
Brazil
E-mail: lauraandrade18@hotmail.com

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