

Do you know this syndrome? *

Você conhece esta síndrome?

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CASE REPORT

One year and four months old female patient presented since she was 6 months old, progressive hair loss in the occipital region and also alteration of the skin color on the abdominal region. Perinatal history could not be questioned as the child had been abandoned by her biological mother immediately after birth. General physical examination showed short stature and malnutrition, microstomia, thin lips, alopecia predominantly in the occipital, temporal and frontal regions, reduction of the eyelashes. There were scleroderma plaques on the abdomen, back and lumbar region (Picture 1).

It was carried out skin biopsy of the abdominal region and histopathology showed atrophic epidermis

with hyalinization of the dermis and subcutaneous fat and reduction of adnexial structures.

Biochemical tests showed increase in triglycerides and total cholesterol 286mg/dl and 230 mg/dl, respectively. It was carried out echocardiogram, X-rays of the hands, the wrists and the chest which did not detect any abnormality.

When the patient was 2 years and 6 months old she developed alopecia universalis becoming evident the prominent veins of the scalp. The clinical and laboratory findings led to a diagnosis of Progeria and since then the patient is having a multidisciplinary follow-up. (Pictures 2, 3, 4).



FIGURE 1: At the age of 16 months. Thinning of hair on the forehead and eyebrows, scleroderma plaques on the abdomen



FIGURE 2: Two and a half years old. Alopecia universalis, microstomia



FIGURE 3: Two and a half years old. Prominent veins on the scalp



FIGURE 4: Two and a half years old. Scleroderma plaques on the abdomen and the chest

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DISCUSSION

Progeria or Hutchinson-Gilford Syndrome is an autosomal dominant disease. It was described for the first time in 1886, by Hutchinson, and ratified by Gilford, in 1904. It occurs sporadically, with an incidence of 1 in 8 million live births and there are approximately 150 cases described in the medical literature. It predominates in males with a ratio of 1,5:1 and it is also observed greater susceptibility of Caucasians in 97% of cases.^{1,2,3,4}

It is characterized by premature aging presenting a rate 7 times higher than the rate of normal aging, causing alterations in many organs and systems, such as the skin, cutaneous tissue, hair cardiovascular and skeletal systems.^{1,4,5}

The genetic basis was discovered in 2003, with findings of mutation in the gene LMNA, which encodes the Blade A generating production of an aberrant protein called progerin, classifying this disease in the group of the so called laminopathies. Progerin is present in high concentration in the cells of these patients promoting distortion in the nuclear membrane, altering the function of chromatin and therefore reducing life expectancy.^{1,3,4,5,6}

Carriers of this disease look normal at birth and the first manifestations of the disease can be seen at the end of their first year of life when the weight gain and the growth curve reduce, the skin becomes sclerodermiform and the first signs of alopecia appear.^{1,2,4,7}

The clinical manifestations are divided into major criteria and signs usually presents itself as follows: the major criteria are a bird-like face (which occurs around 6

months to one year of age), alopecia, prominent veins on the scalp, big eyes, micrognathia, abnormal and slow dentition, pear shaped chest, short clavicles, bow legs (coxa valga), short upper limbs and prominent articulations, low stature and weight with normal bone age, sexual maturation is incomplete, reduction of the adipose tissue and adequate psycho-motor development with normal intelligence.^{1,2,4,6,8}

Diagnosis is essentially clinical with major criteria appearing during the first and second years of life. As differential diagnosis we numbered other syndromes like acrogeria, pangeria, Bloom syndrome, all characterized by premature aging.^{1,2,9}

Prognosis is detrimental to the health of the patient and life expectancy is around 13 years. The main mortality factors are the cardiovascular diseases (75%) like acute myocardial infarction. Despite the advances in cardiovascular surgery, the low survival rate remains due to the high capacity of the disease to reproduce the erythematous plaques.^{2,5,8}

As of now, there are no specific therapeutics and it is directed only to complications. The recent advances in molecular biology with the recognition of the genetic changes might improve the knowledge of aging in human beings.

As the first manifestations are dermatological, 35% of parents seek a dermatologist that should pay more attention in making an early diagnosis like in case presented herewith. □

Abstract: Hutchinson-Gilford Syndrome (Progeria) is a rare autosomal dominant disease characterized by premature aging. It is reported the case of child whose alopecia started at the age of 6 months on the occipital region. The child also presented scleroderma plaques on the abdomen. This syndrome presents alterations in many organs and systems such as the skin and the skeletal and cardiovascular systems. The diagnosis is clinical and there is no treatment for it but recognition is necessary to minimize early atherosclerosis through the control of dyslipidemia. **Keywords:** Aging; Aging, premature; Diagnosis; Progeria

Resumo: A Síndrome de Hutchinson-Gilford (Progeria) é uma rara doença autossômica dominante, caracterizada pelo envelhecimento precoce. Relata-se caso de uma criança, que aos 6 meses iniciou alopecia na região occipital e placas esclerodermiformes no abdome. Esta síndrome apresenta alterações em vários órgãos e sistemas como a pele, esquelético e sistema cardiovascular. O diagnóstico é clínico e não possui tratamento, porém seu reconhecimento é necessário para minimizar a aterosclerose precoce através do controle da dislipidemia. **Palavras-chave:** Diagnóstico; Envelhecimento; Progeria; Senilidade prematura

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