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Severe atopic dermatitis and growth failure associated with a *STAT5B* gene mutation in a 7-month-old infant

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ABSTRACT

STAT5B (signal transducer and activator of transcription 5B) deficiency is a rare autosomal recessive syndrome characterized by severe postnatal growth retardation, atopic dermatitis, and hormonal and immunological abnormalities. We present the case of a 7-month-old male patient with no significant perinatal history who was referred to our clinic for a skin condition that had been present for 5 months, associated with severe growth retardation (low weight for height). After ruling out the most common causes of early-onset atopic dermatitis (such as food allergies, primary immunodeficiencies, and inborn errors of metabolism), genetic testing was performed, leading to a diagnosis of STAT5B deficiency.

Our objective is to present a patient with a very rare disease that has a highly characteristic presentation, thereby aiding in early diagnosis.

Keywords: *STAT5B; atopic dermatitis; growth failure.*

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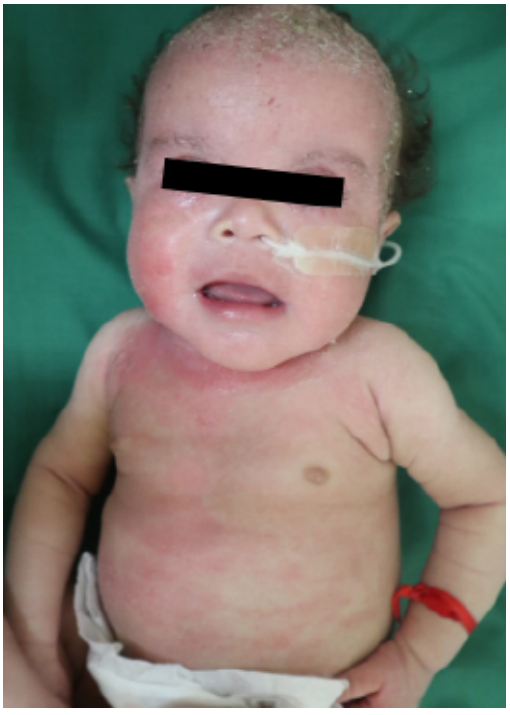
INTRODUCTION

Kofoed *et al*. described STAT5B deficiency (MIM 245590) in 2003 as a rare autosomal recessive syndrome characterized by extreme short stature beginning after birth, chronic eczema, autoimmunity, recurrent infections, and chronic pulmonary complications.[1] The STAT5B pathway is involved in lymphocyte development, survival, and function, as well as in growth.[2] Early recognition of this condition by pediatricians and specialists is of vital importance for preventing the immunological, infectious, and pulmonary complications that lead to death in these patients.

CLINICAL CASE

A 7-month-old male, a full-term newborn with a weight appropriate for his gestational age, was referred to our department due to a skin condition that had been present for 5 months. There was no significant family history, and his personal history included two hospitalizations for gastroenteritis and dehydration associated with poor weight and height growth. On physical examination during the initial consultation, generalized erythematous-desquamative plaques were evident, some of which were exudative, consistent with acute eczema. They were symmetrically located in the cubital, inguinal, popliteal, and neck folds. On the scalp, the patient presented with thick, ichthyosiform scales, sparse, thin, dull hair, and intense pruritus (*Figures 1* and *2*). The patient's facial features were characterized by a depressed nasal bridge and a bulging forehead (*Figure 3*). Regarding anthropometry, the patient was notably short and underweight (below the 3rd percentile for both), a condition that had begun immediately after birth. Neurodevelopmental milestones were within normal limits. Despite poor weight gain, laboratory tests (immunological, nutritional, and general) have consistently remained within normal limits to date. Optical microscopy of the hair shaft revealed no abnormalities, and a skin biopsy performed on an eczematous area in the groin revealed spongiotic dermatitis consistent with atopic dermatitis. Initially, the patient's condition was attributed to an allergy to cow's milk protein, so he went on an elimination diet, but his dermatosis showed no improvement. Moisturizers, topical corticosteroids, and antihistamines were prescribed for his eczema, but there was no response.

Figure 1. Photograph of the head and the trunk



Erythematous-desquamative plaques predominantly affect the trunk and head. Note the appearance of pseudo-macrocephaly due to the prominent forehead and short stature.

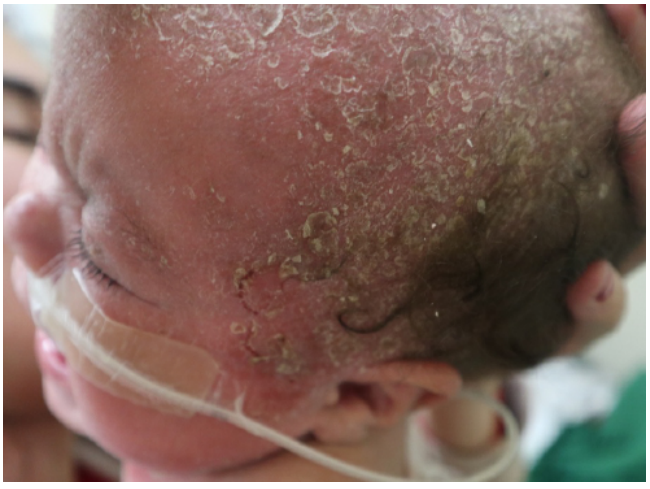
An exome study was conducted to analyze genes associated with ichthyosiform genodermatoses and atopic dermatitis, in which a pathogenic variant was detected in the *STAT5B* gene (NM_012448.4): c.454C>T p.(Arg152Ter), in a homozygous state.

Based on current anthropometric measurements, according to World Health Organization (WHO) guidelines, the child is underweight: 7.9 kg (Z-score: -4.04) and short for age: 70.3 cm (Z-score: -5.79). He is currently 2 years old and remains underweight and short for his age, with no improvement since the initial consultation (*Figure 4*). The patient has experienced respiratory compromise with multiple hospitalizations and respiratory complications, for which he is currently receiving inhaled corticosteroids. Dermatologically, he continues to have severe atopic dermatitis, and a plan is in place to initiate treatment with dupilumab.

DISCUSSION

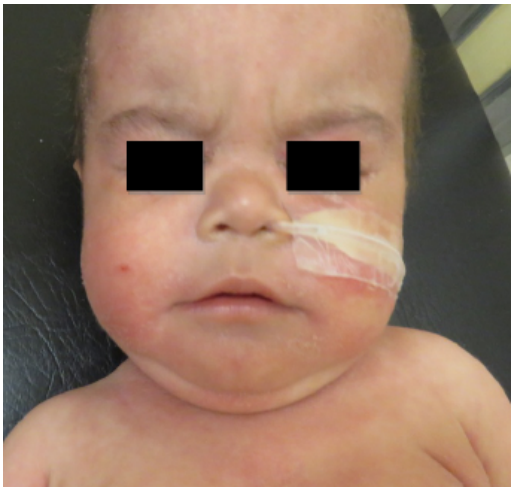
STAT5B deficiency is characterized by immunodeficiency and insensitivity to growth hormone; it is an extremely rare condition, with fewer than 15 cases reported in the literature.[1-3]

Figure 2. Photograph of the scalp



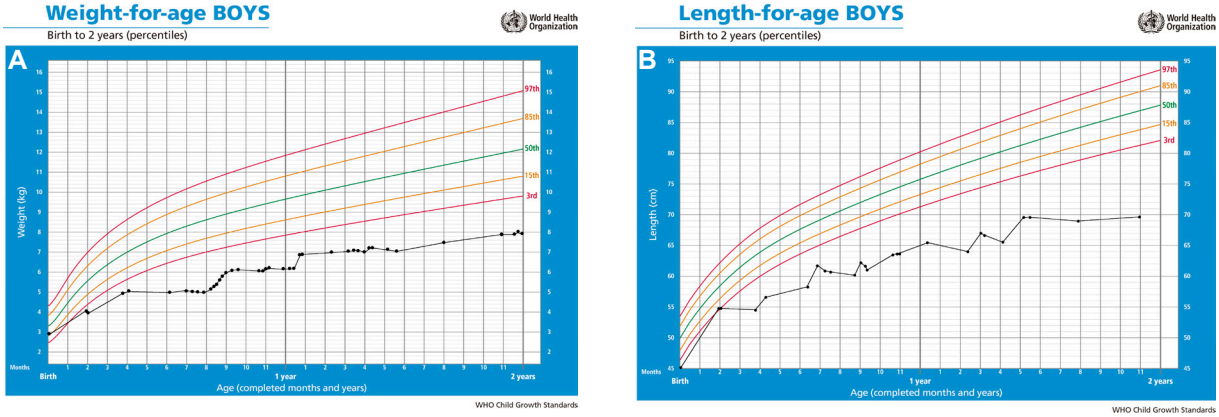
Coarse, ichthyosiform scaling associated with sparse, thin, and dull hair

Figure 3. Frontal view of the face



Depressed nasal bridge and bulging forehead.

Figure 4. Changes in the patient's anthropometric measurements from birth to the present (World Health Organization)



A: Weight-for-age growth chart.
B: Height-for-age growth chart.

Signal transducers and activators of transcription (STAT) are a family of proteins involved in numerous cellular pathways and functions, including the response to cytokines (IL-2, -3, -5, -7, -9, -15, -21) and growth factors, as well as in lymphocyte development and proliferation.[2,4,5]

STAT5B mutations can result in either gain-of-function or loss-of-function effects. Gain-of-function *STAT5B* mutations cause dysregulated allergic inflammation, tumor development, and malignant hematopoietic disorders.[2,4,5] Patients with a loss-of-function *STAT5B* mutation, as in the case of our patient, exhibit impaired postnatal growth because the human growth hormone (HGH) receptor relies on the JAK2-STAT5B signaling pathway to transduce signals from circulating growth hormone to the *IGF1* gene. Furthermore, because STAT is involved in lymphocyte proliferation and interleukin responses, patients with this mutation often experience recurrent infections and immune dysregulation.[2,4]

Phenotypically, patients present with midface hypoplasia, a bulging forehead, a depressed nasal bridge, and a high-pitched voice. The facial features associated with short stature give patients the mistaken impression of macrocephaly. It should be noted that IQ is normal. Birth height is normal for gestational age, with no intrauterine growth restriction; however, after birth, growth deficiency becomes evident (similar to patients with GH deficiency). Puberty is also delayed, reflecting low circulating IGF-1 levels. Additionally, they present severe eczema, as well as an association with autoimmune disorders, such as thyroid disease. Furthermore, they often exhibit progressive immunodeficiency, which compromises the T-cell and NK-cell populations; the prognosis is poor: they die in their fourth decade due to pulmonary disease (alveolar macrophage dysregulation associated with alveolar proteinosis).[3,4] Laboratory findings include elevated prolactin and IgE levels, normal baseline GH levels, but decreased IGF-I levels and T-cell lymphopenia (CD4+ and CD8+) in childhood.[1,4] Furthermore, autoimmune diseases such as juvenile idiopathic arthritis, interstitial lung disease, inflammatory bowel disease, Hashimoto's thyroiditis, and type 1 diabetes have been reported.[2] Although few cases have been described in the literature, most have a poor prognosis; patients die before the age of 30 due to progressive pulmonary fibrosis associated with alveolar proteinosis.[4] Current treatment options are limited and yield variable results. For immune dysregulation, the use of cyclosporine, tacrolimus, sirolimus, and corticosteroids has been described. A lung transplant was indicated in a patient with chronic lung disease and resulted in temporary improvement.[4] Currently, bone marrow transplantation is being considered, while the use of recombinant human IGF-1 therapy remains uncertain, with limited efficacy in promoting growth.[4] A deeper understanding of the mechanisms by which STAT5B regulates function in the hematopoietic system will be key to designing targeted therapies for patients with

this disease.[2] Due to early growth retardation and the key role of STAT5B in GH action, most cases reported in the literature have focused on endocrine and immunological complications. However, pediatricians and dermatologists should consider this condition in children who present severe, early-onset atopic dermatitis associated with characteristic facial features and marked postnatal growth delay.

We emphasize the importance of multidisciplinary follow-up and regular screening to detect immunodeficiencies, autoimmune disorders, and severe lung disease as this condition progresses. ■

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