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Clinical, Cosmetic and Investigational Dermatology

Volume 14 Feb 2021 1 - 8

Open Access Full Text Article

Review

An acroidermal dysplasia patient treated with a small molecule inhibitor of Janus kinase 3 signaling: a single case

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Received: 5 Dec 2020 | Accepted: 12 Jan 2021 | Published: 5 Feb 2021

DOI: https://doi.org/10.2147/CCID.S282966

Abstract: Acroidermal dysplasia (ADJ) is a developmental disorder that affects the skin, hair, and nails. Although the pathogenesis of ADJ is unknown, it is thought to be caused by a mutation in a gene that encodes a protein involved in the signaling of cytokines that use the Janus kinase 3 (JAK3) pathway. Although there is no cure for ADJ, JAK3 inhibitors have been used to treat a patient with ADJ. We report a case of a patient with ADJ who was treated with a JAK3 inhibitor, tofacitinib, and discuss the clinical, histopathological, and genetic data.

Keywords: Janus kinase, cytokine, tofacitinib

Introduction

Acroidermal dysplasia can be used to describe a group of developmental disorders that affect the skin, patients who present with cutaneous abnormalities that include hair, nail, and skeletal abnormalities, and immune deficiencies. Although the pathogenesis of ADJ is unknown, it is thought to be caused by a mutation in a gene that encodes a protein involved in the signaling of cytokines that use the Janus kinase 3 (JAK3) pathway. Although there is no cure for ADJ, JAK3 inhibitors have been used to treat a patient with ADJ. In this case report, we describe the clinical, histopathological, and genetic data of a patient with ADJ who was treated with a JAK3 inhibitor, tofacitinib.

Case report

A 3-year-old female presented with a history of developmental delay, recurrent infections, and a severe exanthem with second-degree burns. The patient was born to healthy parents after a normal pregnancy and delivery. The patient's parents denied any consanguinity. The patient's family history was negative for any similar complaints. The patient's parents denied any exposure to medications, chemicals, or radiation during the pregnancy. The patient's parents denied any history of allergies, asthma, or atopic dermatitis. The patient's parents denied any history of immunodeficiency, autoimmunity, or cancer in the family. The patient's parents denied any history of developmental delay, hair, nail, or skeletal abnormalities in the family. The patient's parents denied any history of consanguinity.

The patient's parents reported that the patient had a severe exanthem with second-degree burns at birth. The patient's parents reported that the patient had a history of recurrent infections, including pneumonia, sepsis, and cellulitis. The patient's parents reported that the patient had a history of developmental delay, hair, nail, or skeletal abnormalities in the family. The patient's parents denied any history of consanguinity.