

Introduction

Autosomal recessive (AR) Hyper IgE syndrome (HIES) due to dedicator of cytokinesis 8 (DOCK 8) mutation is clinically characterized by elevated immunoglobulin E levels, hypereosinophilia, recurrent infections, atopic dermatitis and food allergies. In order to express the phenotype, an individual must have homozygosity or compound heterozygous inheritance. We present the case of a child with a heterozygous variant of uncertain significance (VUS) of the DOCK8 gene identified during the workup for suspected drug reaction with phenotype of AR .

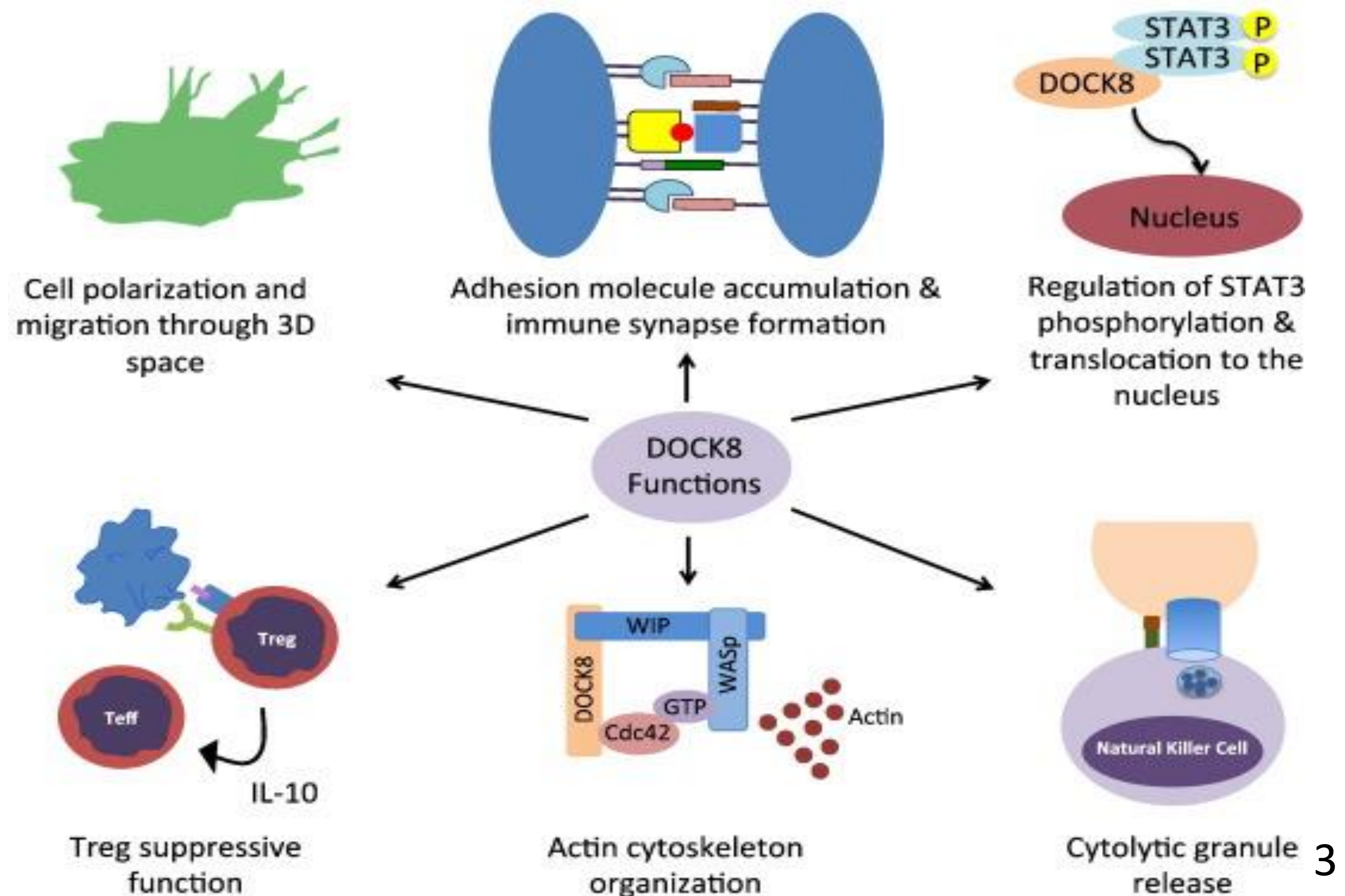
Case Report

The patient is a nine-year-old male with severe poorly controlled eczema with multiple prior skin infections, asthma, allergic rhinitis and multiple food allergies who presented with a diffuse erythematous macular rash, peripheral eosinophilia (absolute eosinophil count 1048) after intravenous antibiotic treatment for a subperiosteal abscess. The patient was referred to Allergy due to clinical concern for Drug rash with eosinophilia and systemic symptoms (DRESS). Further evaluation was notable for significantly elevated total IgE level (11,595 IU/ml) which prompted a workup for possible HIES. Targeted genetic testing showed 5 different VUS mutations. Of these, the discovery of a heterozygous VUS of the DOCK8 gene and his clinical history raised the concern for HIES.

*The opinions or assertions herein are the private views of the authors and are not to be construed as reflecting the views of the Department of the Air Force or the Department of Defense.

Lab Test	Result	Normal
IgE	11595 IU/mL	1.5-144 IU/mL
Absolute eosinophils	1048 cells/uL	30-350 cells/uL
Gene	Patient's Mutation	
DOCK 8	DOCK8 NM_203447.3:c.1241T>G (p.Va1414Gly)	
RANBP2	Chr2(GRCh37):g.109381713G>A.	
HYDIN	Chr16(GRCh37):g.70871718T>A	
SPINK5	Chr5(GRCh37):g.147495989C>T	
ADAMTS13	Chr9(GRCh37):g.136314977C>T	

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2. Izadi N, Leu dermatitis. 10.1016/j.a
3. Biggs CM, K clinical feat 10.1016/j.c PMC555525
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