

5th International Congress on
ALLERGY AND CLINICAL IMMUNOLOGY

May 02, 2022 | Webinar

Received date: 15 November 2021; Accepted date: 18 November, 2021; Published date: 09 May, 2022

Familial Clustering of Juvenile Psoriatic Arthritis associated with a hemizygous FOXP3 mutation

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Purpose of Review: We describe the clinical and genetic findings in four patients from a single family who presented with refractory psoriatic arthritis and were hemizygous in the forkhead box protein 3 (FOXP3) gene (c.1222G>A).

Recent Findings: We report four siblings with hemizygous mutation in the FOXP3 gene (c.1222G>A) who presented with type 1 diabetes mellitus and psoriatic arthritis poorly responsive to treatment. Our findings expand the phenotype spectrum of FOXP3 mutations.

Summary: Immune dysregulation, polyendocrinopathy, and enteropathy, X-linked (IPEX) syndrome is a rare disorder caused by mutations in FOXP3 gene, which lead to early onset of constellation of autoimmune manifestations. This report highlights the influence of immune dysregulation in juvenile arthritis.

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