ESID6-0471
AUTOIMMUNITY, INFLAMMATION AND DYSREGULATION

DOMINATION OF G.380-382DELAA MUTATION AMONG 46 CHILDREN WITH RAG1 DEFICIENCY FROM SOUTH, WEST AND EAST SLAVIC POPULATIONS

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Background:

Mutations in Recombination-activating genes (*RAG*) are common genetic causes of severe combined immunodeficiency (SCID) and Omenn syndrome (OS) and can cause a wide variety of clinical and immunological phenotypes in humans, ranging from absence of T and B-lymphocytes to occurrence of autoimmunity.
Aim- estimate the genetic diversity and clinical presentation of patients with the *RAG1* defects within Slavic populations.

Methods:

We analyzed retrospectively the mutations and the clinical presentation in 46 patients with *RAG1* deficiency from South, West and East Slavic populations.

Results:

The data of 46 patients (20 females, 26 males) from 38 families of Slavic origin (***East***: Russia–11, Belarus – 2, Ukraine–4; ***West***: Poland–11, Czech Republic–4, Slovakia–2; ***South***: Serbia–7, Slovenia–3, Montenegro–1, Croatia–1) was collected. Patients manifested as SCID – 14, as OS – 23 and atypical