PEDIATRIC RHEUMATOLOGY

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CECR1 p.Gly47Arg mutations are not increased in frequency in Turkish Behçet's disease patients compared with healthy controls

B Erer[1*], E Remmers[2], M Takeuchi[2], D Ustek[3], I Tugal-Tutkun[4], E Seyahi[5], Y Ozyazgan[5], A Gul[1], M Ombrello[6], D Kastner[2]

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Question

In Behçet's disease (BD), vasculitis involving blood vessels of nearly all sizes and types may underlie the diverse tissue and organ involvement. Loss of function mutations in the *CECR1* gene (encoding adenosine deaminase 2) have recently been shown to cause a recessive genetic disease, deficiency of adenine deaminase 2 (DADA2). Patients with DADA2 exhibit systemic vasculopathy characterized by intermittent fevers, skin rash, and neurovascular manifestations along with other features that can lead to a diagnosis of polyarteritis nodosa. Patients homozygous for the *CECR1* p.Gly47Arg mutation are reported in two non-consanguineous Turkish families and this mutation is found at low frequency in the Turkish population. We therefore attempted to determine whether some BD cases may be explained by adenosine deaminase 2 deficiency and whether this mutation contributes to BD risk in patients of Turkish ancestry.

Methods

Turkish BD patients (n = 1,609) and controls (n = 1,519) were genotyped for p.Gly47Arg mutations in the *CECR1* gene using a Sequenom assay. The assay interrogated two mutant alleles of the first nucleotide of the Gly47 codon that both encode the glycine to arginine missense change.

Results

We found p.Gly47Arg mutations in 4 BD patients and 3 healthy controls. No individuals (neither cases or controls) carried two mutant alleles. The carrier frequency for p.Gly47Arg mutations was 0.002 in cases and in controls.

Conclusions

These data show that the carrier rate of *CECR1* p.Gly47Arg mutations is very low in Turkish Behcet's patients and not different from controls, suggesting no contribution to Behcet's disease.

Authors' details
[1]Istanbul University Istanbul Medical Faculty, Internal Medicine Division of Rheumatology, Istanbul, Turkey. [2]National Institutes of Health, National Human Genome Research Institute, Bethesda, MD, USA. [3]Istanbul University Institute of Experimental Medicine, Istanbul, Turkey. [4]Istanbul University Istanbul Medical Faculty, Istanbul, Turkey. [5]Istanbul University Cerrahpasa Medical Faculty, Istanbul, Turkey. [6]National Institutes of Health, National Institutes of Arthritis Musculoskeletal and Skin Diseases, Bethesda, MD, USA.

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[1]Istanbul University Istanbul Medical Faculty, Internal Medicine Division of Rheumatology, Istanbul, Turkey
Full list of author information is available at the end of the article