Abstract Preview - Step 3/4

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Title: ***TACI* expression and mutational status in Greek patients with common variable immunodeficiency and selective IgA deficiency**

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Text: **Purpose**: TACI (Transmembrane Activator, calcium modulator and Cyclophilin ligand Interactor) is a transmembrane receptor, which mediates isotype switching in B cells. Recently, mutations in *TNFRSF13B/TACI* gene were found in 10-20% of patients with common variable immunodeficiency (CVID), implying their contribution in the disease pathogenesis and/or phenotypic expression. The purpose of this study was to analyze the mutational status and the expression of *TNFRSF13B/TACI* in Greek patients with CVID and selective IgA deficiency (sIgAD).
Methods: Thirty-two unrelated patients (M/F: 16/17), with a mean age at diagnosis 13.7 years (range: 1-64) were enrolled. Amongst them, 14 suffered from CVID, 13 from sIgAD, 3 from sIgG4D and 2 from transient hypogammaglobulinemia. DNA was extracted from peripheral blood by standard protocol. Amplification of all exons of *TNFRSF13B/TACI* gene was followed and the purified PCR products were directly sequenced. The expression levels of TACI were measured by flow cytometry using an anti-CD267 monoclonal antibody (Abcam, clone: 1a1). 108 healthy controls (M/F: 44/64, mean age: 42.6 years, range: 7-93) were also analyzed for TACI expression and for the presence of the two most common missense polymorphisms (V220A, P251L - detected by PCR-RFLP).
Results: Only the two common *TNFRSF13B* polymorphisms (V220A and P251L) were detected amongst patients, with prevalence, however, that did not differ from that detected in normal controls (allele frequencies V220A: 3.3% vs 4.7%, p=0.79 and P251L: 11.7% vs 10.3%, p=0.6). In immunodeficient patients, B cell TACI expression was unrelated with the presence of *TNFRSF13B/TACI* polymorphisms but its levels were found significantly lower than that detected in normal controls (mean±SDEV: 18.9±20.8 vs 34.8±17.2, p=0.018). In normal controls, the presence of polymorphisms was not associated with hypogammaglobulinemia. In the later group, however, a negative correlation was observed between the P251L polymorphism and TACI expression.
Conclusions: The *TNFRSF13B/TACI* coding region does not seem to dispose disease-associated mutations in Greek patients with primary immunodeficiencies. Differences, however, in TACI expression levels uncovered in this study indicate a possible functional role of common *TNFRSF13B/TACI* polymorphisms that remains to be elucidated.

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