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TITOLO: Genetic analysis of the TNFRSF13B gene in 244 Italian CVID patients addressed to a single centre

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TESTO:

Introduction: Common variable immunodeficiency (CVID) is characterized by hypogammaglobulinemia, recurrent bacterial infections and defective antibody response. Mutations in ICOS, TACI, CD19, CD20, CD21, CD81, BAFF-R, NFKB1, NFKB2, LRBA, ADA2, TWEAK have been associated with CVID; mutations in the gene coding for TACI (TNFRSF13B), are reported to be present in around 8-10% of CVID cases. **Objective.** To screen for mutations in the TNFRSF13B gene in Italian CVID patients, determine the national mutation frequency and compare the results to the ones reported in literature. **Methods.** 244 Italian CVID patients were included in this study. Informed consent was obtained. TNFRSF13B coding regions were amplified through PCR and direct sequencing was performed using an ABI310 sequencer. **Results.** The genetic screening evidenced the presence of 19 known variants, 3 synonymous, 13 missense and 3 nonsense mutations. The frequencies of c.204insA, I87N, C104Y, C104R mutations in our cohort are statistically increased when compared to the general population. Rare mutations (L171R, R72C, R122Q, S144X, R198H, C172Y and C193R) normally not found in the general population, were present in our patients although with low frequencies. Regarding compound heterozygous mutations we identified some already documented ones (C104R/C104Y, c.204insA/C104R) and three novel ones (C104R/L171R; I87N/C104R; R72C/C104Y/R122Q). We also identified 2 mutations not yet reported on the online database (1000G project): Q95X, D191N. **Conclusions.** Our genetic analysis of the TNFRSF13B gene reports new mutations non reported in literature and new compound heterozygous mutations in a large Italian CVID population. The other mutations reported in literature are well represented even if, in some cases, with differences in incidence.

Abstract per il Congresso

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