Group **B** Poster n. **20**

Section n. 6. Genetics of aging

Genetic aspects underlying the Normocalcemic and Hypercalcemic phenotypes of Primary Hyperparathyroidism

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Hypercalcemic primary hyperparathyroidism (PHPT) is a common endocrinedisorder that has been very well characterized. In contrast, many aspectsof normocalcemic primary hyperparathyroidism (NPHPT) such as natural history, organdamage, and management are still matter of debate. In addition, both thepathophysiology and molecular basis of NPHPT are unclear. We investigated whetherPHPT and NPHPT patient cohorts share the same pattern of genetic variation in genesknown to be involved in calcium and/or bone metabolism.
Genotyping for 9 SNPs was performed by Real-Time PCR (TaqMan assays) on 27 NPHPT and 31PHPT patients. The data of both groups werecompared with 54 in house-controls and 503 subjects from the 1,000 GenomesProject. All groups were compared for allele/haplotype frequencies, on a single locus,two loci and multi-locus basis.
Preliminary results showed that the NPHPT group differed significantly at SNPs in OPG and ESR1. Also, the
NPHPT cohort was peculiar for pairwise associations of genotypes and for theoverrepresentation of unusual multilocus genotypes.
In conclusion, our NPHPT patient set harboured a definitely larger quota of geneticdiversity than the other samples. Specific genotypes may help in defining subgroups ofNPHPT patients which deserve ad hoc clinical and follow-up studies. ACKNOWLEDGMENTS: Work supported by POR FESR Lazio 2014-2020 (n. A0375-2020-36631) to P.M. and
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