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Heritability of baseline frequency of micronuclei in buccal cells: the SMP-twin study.

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The analysis of the spontaneous frequency of micronuclei in buccal cells is applied in human biomonitoring studies to quantify genomic damage associated with environmental and occupational exposures, dietary, lifestyle habits as well as clinical outcomes (1-3). It is important to identify the critical factors affecting interindividual variability to evaluate with confidence whether the variations observed among individuals are due to environmental factors or the individual's genetic make-up.

One of the most robust methods for identifying sources of variation in humans is to analyse a specific trait in twins. The comparison of the similarity between pairs of genetically identical twins (monozygotic, MZ) and fraternal twins (dizygotic, DZ) allows to measure the relative contribution of genetics, as opposed to environment, to a given trait.

The design of the present study is aimed at investigating the heritability of baseline frequency of micronuclei in buccal cells from a population of 150 pairs of healthy MZ and DZ twins, enrolled from the Italian Twin Registry, aged from 18 to 80 years. Information on health status, smoking habits and mental well-being status were obtained through questionnaires administered to the enrolled subjects. Biological samples, including buccal cells, serum, plasma and blood mononuclear cells, were appropriately stored in a biobank.

A secondary aim of this study is to determine if the mental well-being status may affect the frequency of micronuclei in buccal cells. A recent study by Reimann et al. (2020) highlighted a possible contribution of depression on DNA damage in buccal cells and suggested the need of further studies to determine whether this analysis could be predictive of mental disorders. In the present study, the analysis of DNA damage will be focused on a subgroup of MZ twins discordant for mental well-being status to better investigate this association with the powerful tool provided by the twin model.

In addition, the measure of micronucleus frequency will be integrated with the analysis of individual levels of biomarkers of oxidative stress and antioxidant capacity to investigate the influence of oxidative stress on this marker of DNA damage.