

Confirmation of deleted Copy Number Variation during aging in monozygotic twins by qPCR

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For many years scientists believed that all cells, originating from a single fertilized egg, were harboring exactly the same genetic information. However, recent evidence confirmed that cells not only have different genetic contents at the beginning but also that the genetic differences accumulate over the years and cause somatic mosaicism. Consequently cells can accumulate gains, deletions, duplications and translocations which in turn can sometimes lead to disease and death.

Nowadays it is believed that aging might be a result of accumulation of these types of changes. Recent studies have shown that single nucleotide polymorphisms, copy number variations and *de novo* mutations can cause somatic mosaicism. Therefore, genomic content may vary across a set of cells in the same individual.

In order to study this phenomenon monozygotic twins provide an ideal model, mainly due to the fact that they share an identical genome at birth and occurrence of any variation over time in one individual can be constantly compared with the other. My project consists of genomic comparison of monozygotic twin pairs. In particular the aim of this project is to explore the genomic changes that could give rise to phenotypic discordance among twins. A particular type of discordance can cause diseases like cancer or an autoimmune disorder that affects one individual of the pair while the other is healthy. The twin cohort used in the study belongs to several projects and includes individuals different for age groups and health status.

The purpose of this work was to validate by wet-lab techniques the changes found in the genome of monozygotic twins by using the whole genome genotyping array platforms acquired from Illumina and Nimblegen.

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