124- A LARGE STUDY ON THE FREQUENCY OF SOMATIC CNV IN LYMPHOCYTES

The human genome project disclosed that up to 5.5% of our genome is composed of segmental duplications. These duplications started, obviously, from a single event which was then fixed in the population. In 2004 two simultaneous papers, in Nat Genet[1] and in Science[2], documented for the first time that Copy Number Variations (CNVs) are indeed present in the human population. This achievement was possible by exploiting the micro-array technology. Next step was the discovery, by the J.P. Dumansky's group[3], that CNVs can discriminate different tissues of the same individual. The possibility of analysing single cells further improved our knowledge of somatic mosaicism for CNV.
In this context, Liu et al., in a paper in Genome Res., have published a large-scale single-cell whole-genome profiling of normal human lymphocytes (20,000 lymphocytes from 16 individuals), allowing a detailed statistics on this topic. 7.5% of the cells had large-size copy number alterations. Trisomy 21 was the most prevalent autosomal aneuploidy. Monosomy X occurred most frequently in females older than 30 years.

1- Iafrate et al.: Detection of large-scale variation in the human genome. Nat Genet 36:949-51 (2004)

2- Sebat et al.: Large-scale copy number polymorphism in the human genome. Science 305:525-528 (2004)

3- Piotrowski et al.: Somatic mosaicism for copy number variation in differentiated human tissues. Hum Mutat 29:1118-1124 (2008)

4- Liu et al.: Low-frequency somatic copy number alterations in normal human lymphocytes revealed by large-scale single-cell whole-genome profiling. Genome Res (2021) *https://genome.cshlp.org/content/early/2021/12/28/gr.275453.121.long*