Trisomy 21

The Genetic Basis for Down Syndrome

95% percent of individuals with Down Syndrome have full Trisomy 21 which is due to a supernumeraray chromosome 21 in every cell of the body. It results due to nondisjunction, or failure of a pair of chormosomes to disjoin during either of the two meiotic divisions, usually in the egg (usually Meiosis I), and in rare occasions in the sperm. The risks for non-disjunction are multifactorial, the two main contributing factors being increased maternal age and altered recombination [1].

In eggs, Meiosis is initiated when the mother herself is still in utero, at about 11–12 weeks of gestation. The process is arrested at in prophase I until just prior to ovulation (for decades!). At this point the oocyte completes Meiosis I and progresses to metaphase II where it remains. Finally, at fertilization the meiotic process is completed. In contrast, spermatogenesis begins at puberty. The cells enter meiosis and progress from one stage to the other with no delay.

The increased in risk for non-disjunction in the female parent is probably because of the protracted phase of meiotic arrest, which probably allows the risk factors to accumulate in the milieu of the ovarian environment [2].

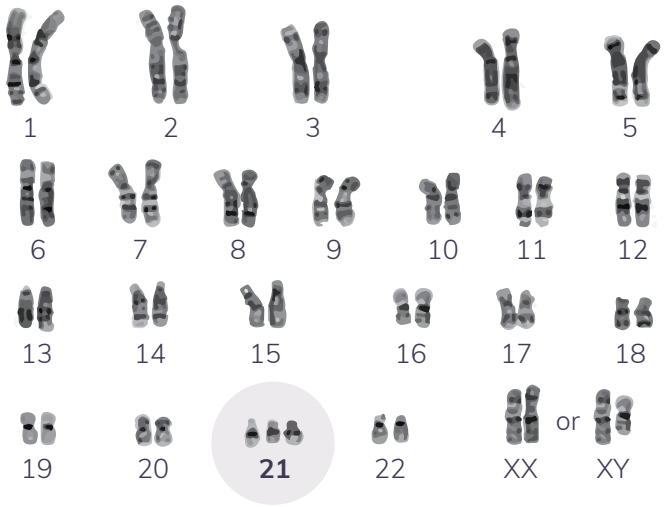

Figure 1: Trisomy 21 Karyotype [3]

Autosomes (1-22)
Sex Chromosomes (XX, XY)

1 Oliver TR, Feingold E, Yu K, Cheung V, Tinker S, Yadav-Shah M, et al. (2008) New Insights into Human Nondisjunction of Chromosome 21 in Oocytes. PLoS Genet 4(3): e1000033. https://doi.org/10.1371/journal.pgen.1000033

2 Sujoy Ghosh, Chang-Sook Hong, Eleanor Feingold, Papiya Ghosh, Priyanka Ghosh, Pranami Bhaumik, Subrata Kumar Dey, Epidemiology of Down Syndrome: New Insight Into the Multidimensional Interactions Among Genetic and Environmental Risk Factors in the Oocyte, American Journal of Epidemiology, Volume 174, Issue 9, 1 November 2011, Pages 1009–1016.

3 Trisomy 21 Karyotype. U.S. National Library of Medicine. https://ghr.nlm.nih.gov/primer/mutationsanddisorders/chromosomalconditions