Mosaicism

down syndrome care hub

The Genetic Basis for Down Syndrome

Some individuals with Down syndrome have an extra copy of chromosome 21 in some, but not all of their cells.

Mosaicism is usually a result of nondisjunction during an early postzygotic mitotic division. For example one cell line may lose the additional chromosome 21. The individual continues development with some cells having 47 chromosomes and some 46. Although it has sometimes been observed that those who are mosaic have subtler characteristics of Down syndrome, it is difficult to assess. Testing any given sample cannot reflect or predict the proportions in other tissues.

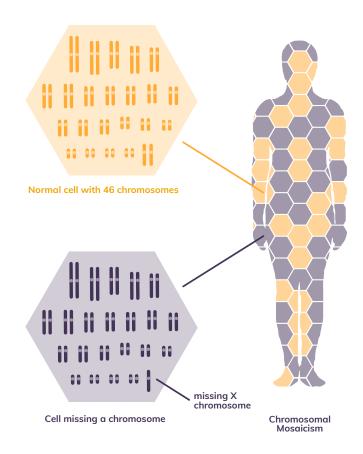


Figure 1: Chromosomal Mosaicism 4

⁴ Chromosomal Mosaicism. U.S. National Library of Medicine. https://ghr.nlm.nih.gov/primer/mutationsanddisorders/chromosomalconditions