

## Phenotypic impact of Robertsonian Translocations on their carriers

Robertsonian translocations (RTs) are detected in roughly 1 per 800 referrals for prenatal diagnostics. While carriers of RTs are thought to harbor themselves no phenotypes, they are at increased risk for miscarriages, infertility, uniparental disomy, and aneuploid offspring because of production of unbalanced gametes. A recent cohort study with a median follow up time of 24 years in the United Kingdom revealed a significantly increased risk for breast cancer for carriers of a rob(13:14). Also the risk for non-Hodgkin lymphoma and childhood leukemia was elevated, albeit that this risk was not related to a specific type of RT. Poot and Hochstenbach reviewed the literature regarding potential mechanisms by which RTs may arise, and the effect of RTs on nucleolar structure in view of their pathological effects in somatic cells ([Molecular Syndromology](#)). The authors also discuss the abilities and limitations of current molecular and cytogenetic methods to detect RTs in a clinical diagnostic setting.