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The laboratory is a pioneer in developing genomic methodologies to understand the causes and consequences of structural genomic variation. Using clinical samples as input, the laboratory studies the mechanisms underlying chromosomal instability and rearrangements and the identification of gene or chromosomal mutations leading to developmental abnormalities. A main focus is understanding the phenotypic and structural variability in 22q11DS. The lab actively develops methods for single cell CNV detection and will continue to address outstanding questions about instability especially during human development using single cell genomic approaches. The genomic methods are translated towards preimplantation, prenatal and postnatal diagnosis. The group is partner of the SymbioSys, the systems biology center of excellence in computational biology. Prof. Vermeesch has published over 300 papers with an H-index 49 and is founder of a spin-off company, Cartagena.