Search for Structural Variants in Patients With DSD and Inconclusive Molecular Diagnosis

NCT05867979

Status	RECRUITING
Phase	Not Applicable
Sponsor	University Hospital, Montpellier
Enrollment	20 participants

Key Eligibility Criteria

Inclusion (3)

- homogeneous XY male karyotype.
- patient at least 6 months old
- severe to moderate DSD (Prader 1 to 5) for which the molecular diagnosis is inconclusive after a gene panel analysis.

Exclusion (3)

- subject with a homogeneous or mosaic XX, or monosomal X karyotype.
- subject with an aneuploidy.
- subject with a conclusive molecular diagnosis explaining the observed DSD (i.e. carrier of a causal genotype already well characterized by functional studies)

Locations (1 total)

University Hospital Montpellier, Montpellier, France