Contribution of Optical Genome Mapping (OGM) in the Diagnosis of Multiple Congenital Malformations With or Without Intellectual Disability Without Genetic Abnormality

NCT07370792

Status	RECRUITING
Phase	Not Applicable
Sponsor	Céline PEBREL-RICHARD
Enrollment	55 participants

Key Eligibility Criteria

Inclusion (8)

- Patients presenting with at least two congenital anomalies, with or without intellectual disability, and weighing more than 5 kg.
- Whole-genome sequencing performed by the AURAGEN laboratory deemed non-contributive (absence of class 4 or 5 variants, or identification of a VUS, or identification of only one variant in the context of a recessive disorder).
- Patient covered by a social security scheme.
- Patient able to understand and to oppose participation in the study.
- Written informed consent for genetic analyses, signed either by the patient or by their legal representatives (for minors), after clear and fair information about the study has been provided.

... and 3 more (see full listing online)

Locations (1 total)

CHU clermont-Ferrand, Clermont-Ferrand, France