

Characterization and Contribution of Genome-wide DNA Methylation (DNA Methylation Episignatures) in Rare Diseases With Prenatal Onset

NCT06475651

Status RECRUITING
Sponsor Assistance Publique - Hôpitaux de Paris
Enrollment 63 participants

Key Eligibility Criteria

Inclusion (14)

- Fetuses with a postmortem examination as part of the etiological diagnosis of developmental abnormality within the Genomic Medicine of Rare Diseases department of the Necker Children's Hospital, and whose DNA extracted from lung and amniotic fluid is available
- OR a child cared for in the Genomic Medicine for Rare Diseases department of the Necker Children's Hospital, and whose DNA extracted from whole blood is available
- with pathogenic or probably pathogenic variation in a gene following CHD7, KMT2D, HYLS1, TCTN3 or FLVCR2
- whose parents have consented to molecular genetic testing as part of diagnosis and research
- Negative Controls :
... and 9 more (see full listing online)

Exclusion (2)

- Refusal of postmortem examination in case of fetal loss
- Parents' refusal of molecular investigations

Locations (1 total)

Department of Genomic Medicine for Rare Diseases and the Multidisciplinary Center for Prenatal Diagnosis of the Necker-Enfants malades Hospital, Paris, France