NIHR | BioResource

PND. Paediatric Neurodevelopmental Disorders

NIHR BioResource – Rare Diseases study project

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Summary

Paediatric neurodevelopmental disorders are a major cause of disability presenting in early life, often with long term consequences requiring considerable family and social support. Recent advances in genomics have shown that most have a genetic basis, and many can be diagnosed with the whole genome and whole exome sequencing. It has proven particularly powerful to detect de novo dominant mutations in trios. Although many of these disorders have no cure, a growing proportion have an identified neurometabolic basis opening new avenues for new therapy development. In addition, international efforts to develop gene therapy approaches have led to clinical trials, led by academia or in partnership with industry.

The BioResource aims to continue to build cohorts of patients with specific paediatric neurodevelopmental disorders. Our emphasis is on diagnosis, understanding the natural history, and developing new treatments.

Recruitment Criteria

We are recruiting patients with a paediatric neurodevelopmental disorder. Although many will have a molecular genetic diagnosis, some will not have a diagnosis. These can be recruited on clinical grounds.

Inclusion

Patients with a clinical diagnosis of neurodevelopmental disorder beginning below the age of 18.

We encourage the recruitment of multiple affected and unaffected members of the same family.
We encourage the recruitment of syndromic and non-syndromic disorders.

Exclusion

Children unlikely to have a genetic cause for their neurodevelopmental disorder such as perinatal injury or a brain tumour or an inflammatory disorder.