

Further Delineation of the De Santo Shinawi Syndrome Phenotype Using a Series of Individuals Carrying a Pathogenic Variant of the WAC Gene

NCT06807723

Status RECRUITING
Sponsor University Hospital, Clermont-Ferrand
Enrollment 50 participants

Key Eligibility Criteria

Inclusion (2)

- Children and adults of any age.
- Molecular diagnosis of a pathogenic (or likely pathogenic) variant involving the WAC gene (SNV, CNV, SV).

Exclusion (3)

- Patients with a molecular diagnosis of another VP (SNV) of a gene responsible for a neurodevelopmental disorder.
- Patient having already participated in a DESSH study with published data.
- No patient data available.

Locations (1 total)

Clermont-Ferrand University Hospital, Clermont-Ferrand, Auvergne, France