Characterization of New Phenotypes of Patients With Spinal Muscular Atrophy Treated With SMN Restoring Therapy

NCT06321965

Status	RECRUITING
Phase	Not Applicable
Sponsor	Hospices Civils de Lyon
Enrollment	60 participants

Key Eligibility Criteria

Inclusion (5)

- Genetically confirmed infantile or juvenile spinal muscular atrophy
- Treated with a therapy that restores SMN protein expression (e.g. nusinersen, risdiplam, onasemnogene abeparvovec)
- Aged 0 to 15 years inclusive
- Informed consent signed by both parent(s)/legal guardian(s) and patient's assent
- Affiliated or beneficiary of a health insurance plan*. * for inclusion in France

Exclusion (4)

- Other condition likely to interfere significantly with ASI assessment and clearly unrelated to the disease
- Other associated neurological disease
- Current pregnancy or breast-feeding (a pregnancy test will also be performed at inclusion).
- Please note that patients with a specific contraindication to MRI (i.e. metallic foreign body, claustrophobia and other reasons determined by the investigators) will be allowed to participate in the study, but MRI will not be performed.

Locations (8 total)

Pediatric Rehabilitation Service - L'Escale Mother and Child Hospital, Bron, Rhone, France
CHRU of Brest, Brest, France
Pediatric Neurology and Resuscitation Raymond-Poincare Hospital, Garche, France
... and 5 more locations