

# Gene Replacement Therapy for Treatment of Paediatric Patients With CTNNB1 Neurodevelopmental Syndrome

NCT07270549

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Status	RECRUITING
Phase	Phase 1, Phase 2
Sponsor	CTNNB1 Foundation
Enrollment	12 participants

## Key Eligibility Criteria

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### Inclusion (13)

- Male or female participant aged 2-12 years at the time of informed consent (Part A: 6-12 years, Part B: 2-12 years).
- Child aged 4 to 12 years has to weigh at least 13,3 kg: 5,0E+14 vg.
- Child aged 3 years has to weigh at least 11,96 kg: 4,5E+14 vg.
- Child aged 2 years has to weigh at least 10,94 kg: 4,11E+14 vg.
- Genetically confirmed diagnosis of CTNNB1 syndrome with a heterozygous pathogenic or likely pathogenic variant in the CTNNB1 gene (Class 4/5 according to American College of Medical Genetics and Genomics), confirmed by geneticist at screening.

... and 8 more (see full listing online)

### Exclusion (44)

- Participant has a mutation in the CTNNB1 gene which is predicted to result in a gain-of-function effect (e.g. p.G575R) or dominant negative effect (e.g. p.Y333\\*, p.Q193\\*, p.A317Vfs8\\* and p.S352fs\\*) on the Wnt/<sup>2</sup>catenin pathway, or any variant that, in the opinion of the PI, is inconsistent with the mechanism of action of the gene replacement therapy.
- Participant has a concomitant genetic diagnosis or neurodevelopmental syndrome that in the opinion of the investigator could interfere with safety, ability to perform assessments, or data interpretation.
- Participant tests positive for AAV9 antibody with titers  $\gt 1:50$  for AAV9 antibodies utilizing an enzyme linked immunospot.
- Participant has a known allergy or hypersensitivity to any ingredients or excipients of the IMP, or to immunosuppressants or pre-medications specified within the trial protocol.
- Participant with a history of receiving immune-modulating agents (such as chemotherapy, radiotherapy, intravenous steroids, other immunosuppressive agents) within 3 months prior to dosing. Topical or inhaled corticosteroid treatment may be permitted at the discretion of the investigator.

... and 39 more (see full listing online)

## Locations (1 total)

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University Medical Centre Ljubljana, Ljubljana, Slovenia

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<https://clinicaltrials.gov/study/NCT07270549>

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