Therapeutics for Phelan-McDermid-Syndrome

Antisense oligonucleotides stabilizing Shank3 mRNA

Technology

Phelan-McDermid syndrome (PMDS) is a rare genetic disorder caused by a deletion or disruption of a small part of chromosome 22, which includes the SHANK3 gene. This syndrome is characterized by developmental delay, intellectual disability, delayed or absent speech, and often features such as autism spectrum disorder, seizures, low muscle tone (hypotonia), and distinctive facial features. So far no known treatments have been successful at targeting the underlying cause of PMDS. Instead, current treatments are based on symptom management and monitoring organ function.

The inventors present a potential novel causative approach to treatment of haploinsufficient PMDS patients, which constitute the major group of patients. Antisense oligos (ASO) directed against the 3' UTR of Shank3 mRNA was shown to increasing Shank3 expression in a cell culture model. Two ASOs were identified that enhanced SHANK3 expression by about 1.3–1.6-fold, which can be expected to be sufficient for successful treatment. The technology is open for licensing or collaboration projects with the responsible scientist.

The technology is open for licensing, as well as for co-development with the inventors.

Innovation

- Novel causative treatment for PMDSVTNC, BGN, H10 high
- Specific antisense oligonucleotides stabilizing Shank3 mRNA

Application

- Major group of Phelan-McDermid Patients
- Heterozygous loss of Shank 3 gene

Responsible Scientist

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Patent Status

Pending Applications
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