ISTITUTO ITALIANO
DI TECNOLOGIA

TITLE

Neuro-specific viral vector for gene therapy to treat cognitive impairment in Down syndrome by RNA interference

INVENTORS

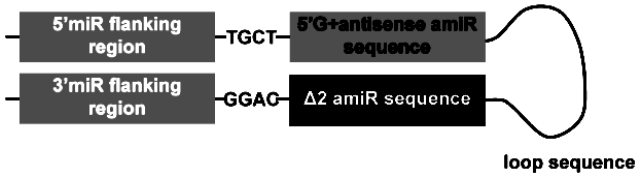
Andrea CONTESTABILE, Laura CANCEDDA, Giuseppe RONZITTI, Federico MINGOZZI

DESCRIPTION

The invention deals with the use of artificial micro RNA (amiR) to reduce the expression of NKCC1 protein (encoded by SLC12A2 gene), which is involved in the pathogenesis of several diseases via regulation of intracellular chloride concentration.

The invention is based on recent studies showing that NKCC1 is overexpressed in the brains of patients with Down Syndrome and pharmacological treatment with the inhibitor Bumetanide restores synaptic plasticity and cognitive impairment in mouse models. However, several data indicated that a life-long treatment with Bumetanide would be required, thus potentially inducing undesirable excessive diuresis and related side effects of chronic imbalance in a number of electrolytes.

The invention therefore provides an engineered system to achieve neuron-specific expression of specific amiR against NKCC1 by using a human Synapsin promoter to drive transgene expression.



APPLICATIONS

Gene therapy, treatment of cognitive impairment due to Down syndrome

KEYWORDS

NKCC1 inhibition, viral vector, microRNA

BIBLIOGRAPHIC DATA

Methods and pharmaceutical composition for reducing the expression of NKCC1 in a subject in need thereof

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Applicants	Fondazione Istituto Italiano di Tecnologia, Institut National de la Santé et de la Recherche Médicale (INSERM), Université d'Evry-Val-d'Essonne, GENETHON

CONTACTS

Technology Transfer Office	Lorenzo Rossi	+39 010 71781 489
		lorenzo.rossi@iit.it