Clinical Trial Protocol
Iranian Registry of Clinical Trials

10 Jun 2026

Survey of Miglustat therapeutic effects on neurological and systemic symptoms of infantile type of Sandhoff and Tay-Sachs diseases

Protocol summary

Study aim

Therapeutic effect of Miglustat on infantile type of Tay-sachs and Sandhoff diseases

Design

Treatment with Miglustat starts for one year. Patients are evaluated at the beginning of treatment and every 4 months. Then after, Miglustat is stopped and patients are followed for 1 year after treatment cessation and compared before and after treatment regards to neurological and systemic outcomes. Although randomized control trial is the gold standard for clinical trial studies; there are ethical concerns about placebo control group in rare diseases such as Sandhoff and Tay sachs diseses.

Settings and conduct

Patients are evaluated for neurologic examination, seizure, feeding with NG-T, aspiration pneumonia and quality of life in frequent visits. Variables in neurologic examination are muscle tone, muscular atrophy and contracture. Motor function is scored according to Gross Motor Function Classification System scoring system (GMFCS) and quality of life is assessed by Infant Toddle Quality Of Life (ITQOL) questionnaire, with confirmed validity and reliability. Secondary outcomes are compared during treatment and after treatment sessation.

Participants/Inclusion and exclusion criteria

Patients who are registered with diagnosis of Sandhoff and Tay sachs, at Myelin Disorders Clinic, Children's Medical Center confirmed by enzyme study and genetic tests. Exclusion criteria: diarrhea (more than 3 times daily), Renal impairment (Cr Clearance <1.73), loss of follow up visits, other systemic diseases, concomitant treatment.

Intervention groups

Treatment with Zavesca regimen based on body surface area as follows: SQRT [Height (cm) × Weight (kg)] / 3600 <1.25 : 200mg TDS 0.88- 1.25: 200mg BID 0.73- 0.88: 100 mg TDS 0.47- 0.73: 100 mg BID <0.47: 100 mg Daily

Main outcome variables

Hospitallization Feeding with NG-T versus oral Seizure frequency Aspiration pneumonia Motor function Quality of life

General information

Reason for update

Acronym

IRCT registration information

IRCT registration number: **IRCT20181023041435N1**
Registration date: **2018-11-04, 1397/08/13**
Registration timing: **prospective**

Last update: **2018-11-04, 1397/08/13**
Update count: **0**

Registration date

2018-11-04, 1397/08/13

Registrant information

Name

Motahare Talebian

Name of organization / entity

Country

Iran (Islamic Republic of)

Phone

+98 21 4425 8510

Email address

mh.69tn@gmail.com

Recruitment status

Recruitment complete

Funding source

Expected recruitment start date

2018-11-22, 1397/09/01

Expected recruitment end date

2020-11-21, 1399/09/01

Actual recruitment start date

empty

Actual recruitment end date
empty
Trial completion date
empty

Scientific title
Survey of Miglustat therapeutic effects on neurological and systemic symptoms of infantile type of Sandhoff and Tay-Sachs diseases

Public title
Effects of Miglustat on systemic and neurological symptoms of Sandhoff and Tay-Sachs diseases
Purpose
Supportive
Inclusion/Exclusion criteria
Inclusion criteria:
Clinically and genetically confirmed cases of Sandhoff (SD)/Tay-Sachs (TSD) diseases Enrolled patients who are referred to Myelin Disorders Clinic, Children's Medical Center with clinically suspicion to TSD and SD
Exclusion criteria:
Diarrhea Renal impairement Loss of follow up Other systemic diseases Concomitant drug therapy which may affect neurological system function
Age
From **6 months** old to **24 months** old
Gender
Both

Phase
4
Groups that have been masked
No information
Sample size
Target sample size: **30**
Randomization (investigator's opinion)
N/A
Randomization description
Blinding (investigator's opinion)
Not blinded
Blinding description
Placebo
Not used
Assignment
Single
Other design features
Help to increase quality of life in affected patients

Secondary Ids

empty

Ethics committees

1

Ethics committee
Name of ethics committee
Vice-Chancellor in Research Affairs-Tehran University of Medical Sciences
Street address
No 62, Keshavarz Blvd, Qarib St, CMC hospital
City
tehran
Province
Tehran
Postal code
1419733151
Approval date
2018-10-20, 1397/07/28
Ethics committee reference number
IR.TUMS.VCR.REC.1397.515

Health conditions studied

1

Description of health condition studied
Sandhoff
ICD-10 code
E75.0
ICD-10 code description
GM2 gangliosidosis

2

Description of health condition studied
Tay-Sachs
ICD-10 code
E75.0
ICD-10 code description
GM2 gangliosidosis

Primary outcomes

1

Description
Hospitallization frequency
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
Checklist

2

Description
Pneumonia aspiration frequency
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
Checklist

3

Description
Seizure Frequency
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
checklist

4

Description
nasogastric tube insertion
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
checklist

5

Description
motor function
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
Gross Motor Function Classification System

6

Description
quality of life
Timepoint
before intervention and 4, 8, 12 months after intervention and 1-year without intervention
Method of measurement
Infant/ Toddler Quality Of Life Quastionnaire

Secondary outcomes

empty

Intervention groups

1

Description
Intervention group: Cases of infantile form of Sandhoff and Tay-Sachs diseases
Category
Treatment - Drugs

Recruitment centers

1

Recruitment center
Name of recruitment center
Children's Medical Center
Full name of responsible person
Reza Shervin Badv
Street address
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cmc.tums@gmail.com
Web page address
http://chmc.tums.ac.ir/

Sponsors / Funding sources

1

Sponsor
Name of organization / entity
Tehran University of Medical Sciences
Full name of responsible person
Mohamad Ali Sahraeiyan
Street address
Keshavarz Blvd, Ghods St., Central Organization of the University, 6th floor, Research and Technology Dept.
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office@sina.tums.ac.ir
Grant name
Grant code / Reference number
Is the source of funding the same sponsor organization/entity?
Yes
Title of funding source
Tehran University of Medical Sciences
Proportion provided by this source
100
Public or private sector
Public
Domestic or foreign origin
Domestic
Category of foreign source of funding
empty
Country of origin
Type of organization providing the funding
Academic

Person responsible for general inquiries

Contact
Name of organization / entity
Tehran University of Medical Sciences
Full name of responsible person
Alireza Tavassoli
Position
Associate Professor
Latest degree
Subspecialist
Other areas of specialty/work
Pediatrics

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Sharing plan

Deidentified Individual Participant Data Set (IPD)
Undecided - It is not yet known if there will be a plan to make this available
Study Protocol
Undecided - It is not yet known if there will be a plan to make this available
Statistical Analysis Plan
Undecided - It is not yet known if there will be a plan to make this available
Informed Consent Form
Undecided - It is not yet known if there will be a plan to make this available
Clinical Study Report
Undecided - It is not yet known if there will be a plan to make this available
Analytic Code
Undecided - It is not yet known if there will be a plan to make this available
Data Dictionary
Undecided - It is not yet known if there will be a plan to make this available