

Clinical Trial Protocol

Iranian Registry of Clinical Trials

17 Sep 2026

The effects of N-Acetyl-L-Leucine on the improvement of symptoms in A patient with Multiple Sulfatase Deficiency

Protocol summary

Study aim

The effects of N-Acetyl-L-Leucine on the improvement of motor symptoms in A patient with Multiple Sulfatase Deficiency

Design

The present study is a phase 2 randomized double-blind crossover clinical trial with parallel groups. A total of 1 patient will be admitted to the study from May 22, 2022. The drug allocation sequence is performed in a simple random method.

Settings and conduct

A patient with multiple sulfatase deficiency who referring to Neurology Clinic of the Ghaem Hospital are enrolled in the study. The volunteer, care providers and statistician are blinded after assignment to intervention. So that, the supplements containers were coded as A and B by a non-researcher person and remained confidential until data analysis. The placebos caplet are similar to the supplements regarding the weight and color.

Participants/Inclusion and exclusion criteria

Inclusion criteria: Patient with a definitive diagnosis of multiple sulfatase deficiency having clinical signs, and has not taken any forbidden drugs. Exclusion criteria: Patient with chronic diarrhea, visual loss, malignancies, insulin-dependent diabetes mellitus, known history of hypersensitivity to the N-Acetyl-Leucine, having severe vision or hearing impairment, having a definite diagnosis of arthritis or other musculoskeletal disorders.

Intervention groups

In treatment group (n=1), N-acetyl-L-leucine caplet is taken orally in subject with Multiple Sulfatase Deficiency for 4 weeks and then after a 4-weeks wash-out period, he/she is crossed over to the alternate regimen. In the control group (n=1), placebo caplet of the same shape, weight and colour is used in patients with Multiple Sulfatase Deficiency for 4 weeks and then after a 4-weeks wash-out period, he/she is crossed over to the alternate regimen.

Main outcome variables

The Scale for Assessment and Rating of Ataxia (SARA) score; Spinocerebellar Ataxia Functional Index (SCAFI)

General information

Reason for update

Acronym

IRCT registration information

IRCT registration number: **IRCT20210413050958N5**

Registration date: **2022-06-08, 1401/03/18**

Registration timing: **registered_while_recruiting**

Last update: **2022-06-08, 1401/03/18**

Update count: **0**

Registration date

2022-06-08, 1401/03/18

Registrant information

Name

Maryam Saberi-Karimian

Name of organization / entity

Country

Iran (Islamic Republic of)

Phone

+98 51 3764 3808

Email address

saberikm@mums.ac.ir

Recruitment status

Recruitment complete

Funding source

Expected recruitment start date

2022-05-31, 1401/03/10

Expected recruitment end date

2022-10-07, 1401/07/15

Actual recruitment start date

empty

Actual recruitment end date

empty

Trial completion date

empty

Scientific title

The effects of N-Acetyl-L-Leucine on the improvement of symptoms in A patient with Multiple Sulfatase Deficiency

Public title

Effect of N-Acetyl-L-Leucine in treatment of Multiple Sulfatase Deficiency

Purpose

Treatment

Inclusion/Exclusion criteria**Inclusion criteria:**

Age over 6 years Patient with a definitive diagnosis of Multiple Sulfatase Deficiency Having clinical signs If the patient is taking any medication, he/she should maintain a constant dose/not change his/her treatment during the study period.

Exclusion criteria:

Have not taken any forbidden drugs (including any variant of N-acetyl-DL-leucine, aminopyridines, Riluzole, gabapentin, Varenicline, Chlorzoxazone, sulfasalazine, Rosuvastatin at least 4 weeks before visit 1 and throughout the duration of the study Patient who has any of the following: Chronic diarrhea, Unexplained visual loss, Malignancies, Insulin-dependent diabetes mellitus, Known history of hypersensitivity to the N-Acetyl-Leucine (DL-, L-, D-) or derivatives, History of known hypersensitivity to excipients of Ora-Blend® (namely sucrose, sorbitol, cellulose, carboxymethylcellulose, xanthan gum, carrageenan, dimethicone, methylparaben, and potassium sorbate) Having severe vision or hearing impairment that interferes with their ability to complete study assessments Having a definite diagnosis of arthritis or other musculoskeletal disorders that affects patient's mobility and interferes with their ability to complete study assessments

Age

From 6 years old

Gender

Both

Phase

2

Groups that have been masked

- Participant
- Care provider
- Investigator
- Data analyser

Sample size

Target sample size: 1

Randomization (investigator's opinion)

Randomized

Randomization description

The drug allocation sequence is made in a simple random method. Sequentially numbered sealed envelopes are used to implement the random allocation sequence which opened by a person not involved in the project.

Blinding (investigator's opinion)

Double blinded

Blinding description

The volunteer, care provider and statistician are blinded after assignment to intervention. So that, the supplements containers were coded as A and B by a non-researcher person and remained confidential until data analysis. The placebos are similar to the supplements regarding the weight and color.

Placebo

Used

Assignment

Crossover

Other design features**Secondary Ids**

empty

Ethics committees**1****Ethics committee****Name of ethics committee**

Ethics committee of Mashhad University of Medical Sciences

Street address

Daneshgah Ave., Ghoreishi buildings

City

Mashhad

Province

Razavi Khorasan

Postal code

99199-91766

Approval date

2022-05-17, 1401/02/27

Ethics committee reference number

IR.MUMS.MEDICAL.REC.1401.127

Health conditions studied**1****Description of health condition studied**

Multiple sulfatase deficiency

ICD-10 code

E75

ICD-10 code description

Disorders of sphingolipid metabolism and other lipid storage disorders

Primary outcomes**1****Description**

Movement signs

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Using the Scale for Assessment and Rating of Ataxia (SARA) score and Spinocerebellar Ataxia Functional Index

(SCAFI)

Secondary outcomes

1

Description

The quality of life

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Using PedsQL questionnaire

2

Description

Cell blood count

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Sysmex Cell Counter

3

Description

Lactate dehydrogenase

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

4

Description

Aspartate aminotransferase

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

5

Description

Alanine aminotransferase

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

6

Description

Urea

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

7

Description

Creatinine

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

8

Description

Alkaline phosphatase

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

9

Description

Na

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

10

Description

k

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

11

Description

Total bilirubin

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

12

Description

Direct bilirubin

Timepoint

Before the intervention and 4 weeks after taking supplement or placebo in every study stage

Method of measurement

Auto analyzer instrument

Intervention groups

1

Description

Subject in the intervention group receives N-Acetyl-L-Leucine caplets (daily intake of 2-4 gr depending on the subject's weight) for 4 weeks (n=1) and then after a 4-weeks wash-out period, he/she is crossed over to the placebo. The participant takes the supplement every day, which is contained in an unlabeled bottle.

Supplements are from Hubei ipure Biotech co., ltd (Shenzhen, China).

Category

Treatment - Drugs

2

Description

The subject receives a placebo caplet (daily consumption between 2 to 4 grams depending on the subject's weight) for 4 weeks (n=1) and then after a 4-weeks wash-out period, he/she is crossed over to the alternate regimen (N-Acetyl-L-Leucine). Participant takes a placebo every day orally in an unlabeled bottle. The placebo is prepared by from faculty of pharmacy (Mashhad, Iran) company.

Category

Placebo

Recruitment centers

1

Recruitment center

Name of recruitment center

Ghaem hospital

Full name of responsible person

Maryam Saberi-Karimian

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Ahmadabad Ave.

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Sponsors / Funding sources

1

Sponsor

Name of organization / entity

Mashhad University of Medical Sciences

Full name of responsible person

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Grant name

Grant code / Reference number

Is the source of funding the same sponsor organization/entity?

Yes

Title of funding source

Mashhad 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

Mashhad University of Medical Sciences

Full name of responsible person

Maryam Saberi-Karimian

Position

Associate professor

Latest degree

Ph.D.

Other areas of specialty/work

Biochemistry

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Person responsible for updating data

Contact

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Sharing plan

Deidentified Individual Participant Data Set (IPD)

No - There is not a plan to make this available

Justification/reason for indecision/not sharing IPD

Following a reasonable request, deidentified data will be shared.

Study Protocol

Yes - There is a plan to make this available

Statistical Analysis Plan

No - There is not 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

Yes - There is a plan to make this available

Analytic Code

No - There is not a plan to make this available

Data Dictionary

No - There is not a plan to make this available

Title and more details about the data/document

-

When the data will become available and for how long

-

To whom data/document is available

-

Under which criteria data/document could be used

-

From where data/document is obtainable

-

What processes are involved for a request to access data/document

-

Comments