Clinical Trial Protocol
Iranian Registry of Clinical Trials
13 Dec 2022

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)
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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
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, Chloroxazone, 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

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
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
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).

Description
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
- Treatment - Drugs
- 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
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Name of organization / entity
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Person responsible for general inquiries

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Position
Associate professor

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Ph.D.

Other areas of specialty/work
Biochemistry

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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