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
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Name of organization / entity
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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

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

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).

Category
Treatment - Drugs

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

Recruitment center
Name of recruitment center
Ghaem hospital
Full name of responsible person
Maryam Saberi-Karimian
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Sponsors / Funding sources

Sponsor
Name of organization / entity
Mashhad University of Medical Sciences
Full name of responsible person
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Email
ghayourm@mums.ac.ir

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

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
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Person responsible for scientific inquiries

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**Position**  
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**Latest degree**  
Ph.D.

**Other areas of specialty/work**  
Biochemistry

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Ghaem Hospital, Ahmadabad Ave.

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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**