Clinical Trial Protocol
Iranian Registry of Clinical Trials

30 May 2026

The effects of N-Acetyl-l-Leucine on the improvement of symptoms in patients with ataxia-telangiectasia: A double-blind crossover trial

Protocol summary

Study aim
To determine the effects of N-Acetyl-l-Leucine on the improvement of symptoms in patients with ataxia-telangiectasia

Design
The current study is a randomized, double-blind, crossover clinical trial with parallel groups. A total of 16 subjects are enrolled between Dec.6. 2021 and Aug.6.2022. The table of random numbers was used for randomization.

Settings and conduct
Patients with ataxia-telangiectasia who referring to Neurology Clinic of the Ghaem Hospital are enrolled in the study. All volunteers, 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 caplets are similar to the supplements regarding the weight and color.

Participants/Inclusion and exclusion criteria
Inclusion criteria: Patients with a definitive diagnosis of ataxia telangiectasia having clinical signs and not having addiction, the patients should be on a stable dose/duration and type of speech therapy or physiotherapy. Exclusion criteria: Patients having 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=8), N-acetyl-L-leucine caplet is taken orally in subjects with ataxia-telangiectasia for 6 weeks and then after a 4-weeks wash-out period, they were crossed over to the alternate regimen. In the control group (n=8), placebo caplet of the same shape, weight and colour is used in patients with ataxia-telangiectasia for 6 weeks and then after a 4-weeks wash-out period, they were crossed over to the alternate regimen.

Main outcome variables
Movement signs

General information

Reason for update
-Due to prepare the caplets and not taking sachets, the powder has changed to caplet. -Moreover, due to the unwanted delay in the custom release of the drug, the patients recruitment was extended. -Also, according to a valid evidence in Europe, the supplement dose was adjusted from 1 to 6 grams more precisely to 1 to 4 grams, depending on the patient's weight. -Food recall will also be recorded in patients at each stage of the study.

Acronym

IRCT registration information
IRCT registration number: **IRCT20210413050958N1**
Registration date: **2021-10-27, 1400/08/05**
Registration timing: **prospective**

Last update: **2021-11-25, 1400/09/04**
Update count: **1**

Registration date
2021-10-27, 1400/08/05

Registrant information

Name
Maryam Saberi-Karimian

Name of organization / entity

Country
Iran (Islamic Republic of)

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+98 51 3764 3808

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Recruitment status
Recruitment complete

Funding source

Expected recruitment start date
2021-12-06, 1400/09/15
Expected recruitment end date
2022-08-06, 1401/05/15
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 patients with ataxia-telangiectasia: A double-blind crossover trial

Public title
Effect of N-Acetyl-l-Leucine in treatment of ataxia-telangiectasia
Purpose
Treatment
Inclusion/Exclusion criteria
Inclusion criteria:
Signed informed consent form by the subjects or their parents after explaining the study objectives by the research team Patients with a definitive diagnosis of AT Having clinical signs Not addiction to Drugs and alcohol If the patient is receiving concomitant speech therapy or physiotherapy, he/she has been on a stable dose/duration and type of therapy for at least 4 weeks before visit 1 and throughout the duration of the study 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 Asymptomatic patients Patient who have clinical signs of A-T, but do not have a confirmed genetic test for A-T Patients who have 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 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
No age limit
Gender
Both

Phase
2
Groups that have been masked

- Participant
- Care provider
- Investigator
- Data analyser
- Data and Safety Monitoring Board

Sample size
Target sample size: **16**
Randomization (investigator's opinion)
Randomized
Randomization description
Regarding the presence of the gender as a confounder, we run the block randomization method within each category (male and female) separately. For this reason, inside each category, blocks with size 4 and 2 of the combination of letters A and B (ABBA, ABAB, AABB, BAAB, BBAA, BABA), (AB and BA) are selected to the required number using a table of random numbers and individuals are assigned to groups according to the created sequence.
Blinding (investigator's opinion)
Triple blinded
Blinding description
The random allocation sequence is made using the table of random numbers. Sequentially numbered sealed envelopes are used to implement the random allocation sequence which opened by a person not involved in the project. The participants, care providers and statistician are blinded after assignment to intervention. So that, the caplet bottles are coded by a non-researcher person and remain confidential until data analysis. Moreover, In addition, placebo caplet is similar to supplement ones in shape, 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
2021-08-24, 1400/06/02
Ethics committee reference number
IR.MUMS.MEDICAL.REC.1400.389

Health conditions studied

1

Description of health condition studied
Ataxia-Telangiectasia
ICD-10 code
G11.3
ICD-10 code description
Cerebellar ataxia with defective DNA repair

Primary outcomes

1

Description
Movement signs
Timepoint
Before the intervention and 6 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 6 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 6 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 6 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 6 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 6 weeks after taking supplement or placebo in every study stage
Method of measurement
Auto analyzer instrument

6

Description
Urea
Timepoint
Before the intervention and 6 weeks after taking supplement or placebo in every study stage
Method of measurement
Auto analyzer instrument

7

Description
Creatinine
Timepoint
Before the intervention and 6 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 6 weeks after taking supplement or placebo in every study stage
Method of measurement
Auto analyzer instrument

9

Description
Na
Timepoint
Before the intervention and 6 weeks after taking supplement or placebo in every study stage
Method of measurement
Auto analyzer instrument

10

Description
k
Timepoint
Before the intervention and 6 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 6 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 6 weeks after taking supplement or placebo in every study stage
Method of measurement
Auto analyzer instrument

13

Description
Food recall
Timepoint
Before the intervention and 6 weeks after taking supplement or placebo in every study stage
Method of measurement
24-hour food recall tool

Intervention groups

1

Description
Intervention group: Subjects in the intervention group receive N-Acetyl-L-Leucine caplets (daily intake of 1-4 gr depending on the subjects' weight) for 6 weeks (n=8) and then after a 4-weeks wash-out period, they were crossed over to the alternate regimen. The participants take the supplement every day, which was contained in an unlabeled bottle. Supplements are from Hubei ipure Biotech co., ltd (Shenzhen, China).
Category
Treatment - Drugs

2

Description
Control group: The control group received the placebo (daily consumption between 1 to 4 grams depending on the subject's weight) for 6 weeks (n=8) and then after a 4-weeks wash-out period, they were crossed over to the alternate regimen. Participants take 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
Street address
Ahmadabad Ave.
City
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Province
Razavi Khorasan
Postal code
99199-91766
Phone
+98 51 3840 0001
Email
maryamsabery2012@gmail.com

Sponsors / Funding sources

1

Sponsor
Name of organization / entity
Mashhad University of Medical Sciences
Full name of responsible person
Majid Ghayour-Mobarhan
Street address
Research Council, Ghoreishi bilding, Daneshgah Street
City
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Razavi Khorasan
Postal code
9138813944
Phone
+98 51 3841 1538
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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
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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Ghaem Hospital, Ahmadabad Ave.

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Full name of responsible person
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Position
Associate professor

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Other areas of specialty/work
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Person responsible for updating data

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Full name of responsible person
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Position
Associate professor

Latest degree
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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
Raw data will be shared upon a reasonable request from the corresponding author.

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
Following a reasonable request, deidentified data will be shared.

When the data will become available and for how long
After publication of paper(s) upon a reasonable request

To whom data/document is available
Study PI and executive team

Under which criteria data/document could be used
For reasonable research or clinical purpose

From where data/document is obtainable
Maryam Saberi-Karimian

What processes are involved for a request to access data/document
Direct e-mail

Comments
Moreover, if necessary, other unforeseen genetic, immunologic, biochemical and nutritional assays will be performed in the future only for research purposes on frozen samples.