

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)

Phone

+98 51 3764 3808

Email address

saberikm@mums.ac.ir

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.

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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

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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

Street address

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Person responsible for scientific inquiries

Contact

Name of organization / entity

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Full name of responsible person

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Position

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Latest degree

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Person responsible for updating data

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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.