ForPatients
by Roche

Huntington Disease (HD)

Frequency of Genetic Variations (Selected Single Nucleotide Polymorphisms) in Huntington Disease Gene Expansion Carriers

Frequency of Selected Single Nucleotide Polymorphisms in Huntington Disease Gene Expansion Carriers

Trial Status	Trial Runs In	Trial Identifier
Recruiting	11 Countries	NCT06667414 WE45491

The information is taken directly from public registry websites such as ClinicalTrials.gov, EuClinicalTrials.eu, ISRCTN.com, etc., and has not been edited.

Official Title:

Frequency of Selected Single Nucleotide Polymorphisms in Phase With the Mutant and Wild-Type HTT Alleles in Huntington Disease Gene Expansion Carriers

Trial Summary:

For participation in this epidemiological study, a single-day visit at the study site is required. Participants will be recruited from Huntington Disease clinics, and they will be asked to answer questions regarding their demographics, including sex, age, race and ethnicity, and their medical and medication history. At the end of the visit, a blood sample will be drawn to allow testing with a sequencing assay that is specifically designed for phasing single nucleotide polymorphisms (SNPs) on the wild-type Huntington (wtHTT) and mutant Huntington (mHTT) alleles.

Hoffmann-La Roche	N/A
Sponsor	Phase

NCT06667414 WE45491
Trial Identifiers

Eligibility Criteria:

Gender	Age	Healthy Volunteers
All	#25 Years & # 60 Years	No

1. Why is this study needed?

Huntington’s disease (HD) is a genetic disease caused by a mutated version of the huntingtin gene. HD leads to progressive damage of nerve cells in the brain, which may affect a person’s ability to think, talk, and move. There is a high medical need to develop therapies that treat HD. Understanding more about genetic variations involved in HD may help drug developers direct research efforts.

This epidemiological study aims to find out what types of genetic differences, called single nucleotide polymorphisms (SNPs), are present in the abnormal huntingtin gene and if these genetic differences are more common in some parts of the world. It is common for people to have genetic differences such as SNPs. SNPs do not directly determine the severity or progression of HD. They can be used as a targeting mechanism to guide a selective investigational medicine. This study will help us to better understand HD and how common a certain SNP exists around the world, which will help determine if future clinical trials targeting the SNP may be possible.

2. Who can take part in the study?

People (males / females) of 25 to 60 years of age can take part in the study if they carry the HD gene expansion. Additionally, they have subtle or early symptoms of HD, or are experiencing mild functional impairment.

3. How does this study work?

People will be screened to check if they are able to participate in the study. If eligible, they will be asked to answer a few questions about themselves and their health and to provide a blood sample. The total time of participation in this study will be less than 1 day.

4. What are the main results measured in this study?

The main results obtained in the study are how common it is for people in different parts of the world to have a specific SNP linked to HD.

5. Are there any risks or benefits in taking part in this study?

There is no treatment in this study. Taking part in the study will not make participants feel better. But the information collected in the study can help other people with similar health conditions in the future. Participants in this study may be told of future HD clinical trials that may be of interest to them.

Risks associated with the study procedure

Participants will have their blood drawn one time in this study. They may have unwanted effects of the blood sample collection. Drawing blood may cause pain, bruising, or infection where the needle is inserted. Some people may experience dizziness, fainting, or upset

stomach when their blood is drawn. These unwanted effects can vary from person to person.

Inclusion Criteria:

- Have signed the Informed Consent Form (ICF)
- Aged 25 to 60 years, inclusive, at the time of signing the ICF
- Confirmation of Huntington Disease (HD) gene expansion mutation carrier status
- Confirmation of Total Functional Capacity (TFC) #9 and Total Motor Score (TMS) >6 within 12 months prior to signing the ICF
- Ability to tolerate blood draws

Exclusion Criteria:

- None