

Genetic variation in patients with cerebral palsy

Submission date 06/06/2017	Recruitment status No longer recruiting	☐ Prospectively registered
		☐ Protocol
Registration date 07/06/2017	Overall study status Completed	☐ Statistical analysis plan
		☐ Results
Last Edited 07/06/2017	Condition category Nervous System Diseases	☐ Individual participant data
		☐ Record updated in last year

Plain English summary of protocol

Background and study aims:

Cerebral Palsy (CP) is a term for a number of conditions that affect movement and co-ordination. It occurs when there is a problem in the parts of the brain responsible for controlling muscles. This can be due to abnormal development of the brain or damage caused before, during or after birth. CP leads to a range of symptoms, including muscle stiffness or weakness, random and uncontrolled body movements and balance and coordination problems. Muscle contractures are a common complication of CP, which involve constriction (shortening) in the connective tissues of the body, leading to loss of strength, muscle wasting, pain and rapid fatigue (extreme tiredness). In addition, they cause joints to become fixed in awkward positions, further limiting movement. Patients with muscle contractures often go through numerous extensive operations for joint corrections. The aim of this study is to look at the genes of patients with CP in order to see if there is a genetic explanation for muscle contractures.

Who can participate?

Patients with cerebral palsy and healthy volunteers of the same age.

What does the study involve?

28 patients with CP and four healthy volunteers provide a blood sample which is then used for genetic testing in the laboratory. In addition, 16 CP patients who are having surgery also have sample of tissue collected for further genetic testing.

What are the possible benefits and risks of participating?

There are no direct benefits or risks involved with participating.

Where is the study run from?

1. University of Copenhagen (Denmark)
2. Helene Elsass Center (Denmark)
3. Hvidovre Hospital (Denmark)

When is study starting and how long is it expected to run for?

December 2012 to April 2015

Who is funding the study?

1. Danish Research Council (Denmark)
2. The Elsass Foundation (Denmark)

Who is the main contact?

Dr Jessica Pingel
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Contact information

Type(s)

Scientific

Contact name

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Additional identifiers

Protocol serial number

01

Study information

Scientific Title

Sequence variants in muscle tissue related genes may determine the severity of muscle contractures in cerebral palsy

Study objectives

Cerebral palsy patients show differences in genetic variants in targets that are involved in the structure and metabolism of skeletal muscle tissue.

Ethics approval required

Old ethics approval format

Ethics approval(s)

Regional Ethics Committee for Copenhagen, 30/04/2015, ref: H-4-2014-047

Study design

Observational case-control study

Primary study design

Observational

Study type(s)

Other

Health condition(s) or problem(s) studied

Cerebral palsy

Interventions

Blood samples are taken from 28 individuals with cerebral palsy (CP) and four healthy control reference participants. DNA is extracted from anticoagulated whole blood using the QIAamp DNA Mini Kit (Qiagen, Germany).

Furthermore, tissue samples are taken from 16 patients with CP during surgery and the DNA is extracted from muscle tissue of the medial gastrocnemius muscle using the EZ1 DNA Investigator Kit (Qiagen, Germany).

Intervention Type

Other

Primary outcome(s)

Genetic variants of 96 candidate genes are measured using next generation sequencing and then aligned to a human reference gene (hg19) following the study visit.

Key secondary outcome(s)

Gross motor function is assessed through collection of Gross Motor Function Classification System (GMFCS) scores at the study visit.

Completion date

27/04/2015

Eligibility**Key inclusion criteria**

Cerebral palsy (CP) group:

1. Patients with cerebral palsy in all severity of CP
2. The patient is either over 18 years of age or
3. The patient's parents are legitimate

Healthy participants:

1. Healthy subjects
2. The test person is over 18 years old and authoritative
3. The subjects have no previous musculoskeletal disorders

Participant type(s)

Mixed

Healthy volunteers allowed

No

Age group

Mixed

Lower age limit

18 years

Sex

All

Key exclusion criteria

Cerebral palsy (CP) group:
Individuals with CP without contractures.

Healthy participants:

1. Previous brain injuries
2. Skeletal muscle diseases

Date of first enrolment

01/12/2015

Date of final enrolment

27/04/2016

Locations**Countries of recruitment**

Denmark

Study participating centre**University of Copenhagen**

Department of Neuroscience and Pharmacology
Faculty of Health and Medical Sciences
Blegdamsvej 3.33.3
Copenhagen
Denmark
2200

Study participating centre**Helene Elsass Center**

Holmegårdsvej 28
Charlottenlund
Denmark
2920

Study participating centre
Hvidovre Hospital
Kettegård Alle 30
Hvidovre
Denmark
2650

Sponsor information

Organisation
University of Copenhagen

ROR
<https://ror.org/035b05819>

Funder(s)

Funder type
Research council

Funder Name
Danish Research Council

Funder Name
The Elsass Foundation

Results and Publications

Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are/will be available upon request from Dr. Jessica Pingel (jpingel@sund.ku.dk or jessica.pingel@gmail.com)

IPD sharing plan summary

Available on request