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Genetic variation in patients with cerebral palsy

Submission date	Recruitment status	[ ] Prospectively registered
06/06/2017	No longer recruiting	[ ] Protocol
Registration date	**Overall study status**	[ ] Statistical analysis plan
07/06/2017	Completed	[ ] Results
Last Edited	**Condition category**	[ ] Individual participant data
07/06/2017	Nervous System Diseases	[ ] 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

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Additional identifiers

EudraCT/CTIS number

IRAS number

ClinicalTrials.gov number

Secondary identifying numbers
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

Secondary study design
Case-control study

Study setting(s)
Hospital

Study type(s)
Other

Participant information sheet

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 measure
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.

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

Overall study start date
01/12/2014

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

Age group
Mixed

Lower age limit
18 Years

Sex
Both

Target number of participants
Alltogether 48 participants were recruited for this study

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

Sponsor details
Blegdamsvej 3.33.3
Copenhagen N
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2200

Sponsor type
University/education

Website
www.ku.dk

ROR
https://ror.org/035b05819

Funder(s)

Funder type
Research council

Funder Name
Danish Research Council

Funder Name
The Elsass Foundation

Results and Publications

Publication and dissemination plan
Planned publication in the BMC genomics journal.

Intention to publish date
01/07/2017

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