

# Genetic variation in patients with cerebral palsy

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		<input type="checkbox"/> Protocol
<b>Registration date</b> 07/06/2017	<b>Overall study status</b> Completed	<input type="checkbox"/> Statistical analysis plan
		<input type="checkbox"/> Results
<b>Last Edited</b> 07/06/2017	<b>Condition category</b> Nervous System Diseases	<input type="checkbox"/> Individual participant data
		<input type="checkbox"/> 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  
jpingel@sund.ku.dk

## Contact information

### Type(s)

Scientific

### Contact name

Dr Jessica Pingel

### ORCID ID

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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](mailto:jpingel@sund.ku.dk) or [jessica.pingel@gmail.com](mailto:jessica.pingel@gmail.com))

### **IPD sharing plan summary**

Available on request