

# Brain MR imaging in paroxysmal kinesigenic dyskinesia

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The purpose of this research project is to assess whether affected PKD patients have brain abnormalities compared to healthy normal controls. Hypothesis: Paroxysmal kinesigenic dyskinesia affected individuals exhibit abnormal spontaneous activity of...

<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruiting
<b>Health condition type</b>	Neurological disorders NEC
<b>Study type</b>	Observational invasive

## Summary

### ID

NL-OMON35798

### Source

ToetsingOnline

### Brief title

MRI of the brains in PKD

### Condition

- Neurological disorders NEC

### Synonym

Brief and frequent sudden attacks of involuntary movements

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Erasmus MC, Universitair Medisch Centrum Rotterdam

**Source(s) of monetary or material Support:** Ministerie van OC&W

## Intervention

**Keyword:** Brain, MRI, Neurology, paroxysmal kinesigenic dyskinesia

## Outcome measures

### Primary outcome

The study purpose is to objectify activation patterns in basal ganglia and cortical sensorimotor areas in PKD affected individuals and in healthy normal controls.

Outcome measures:

- Structural imaging: grey and white matter volumes (mm<sup>3</sup>);
- Diffusion tensor imaging: mean diffusivity (MD) and fractional anisotropy (FA);
- Functional imaging: number of significantly ( $P_{corrected} < 0.05$ ) activated voxels;
- Perfusion imaging: cerebral blood flow (ml/s/100 mm<sup>3</sup>).

### Secondary outcome

nvt

## Study description

### Background summary

Paroxysmal kinesigenic dyskinesia (PKD) is a rare neurologic disorder characterized by recurring brief attacks of involuntary movements. PKD is also known as paroxysmal kinesigenic choreoathetosis (PKC, Mount and Reback 1940) [1]. Paroxysmal kinesigenic attacks are initiated by sudden onset moves or startle and can occur up to hundred times a day. The precise phenotype remains unclear and no causative gene is yet found.

PKD belongs to a group of movement disorders characterised by intermittent,

painless attacks of involuntary movements characterised by dystonia, chorea, athetosis and/or ballism. Several classifications have been suggested based on their mode of triggering, duration, frequency and associated syndromes. Kertesz (1967) differentiated a kinesigenic and a non-kinesigenic form [2]. Later, Lance (1977) classified three forms of paroxysmal dyskinesia, according to the duration of the attacks and Demirkiran and Jankovic (1995) described four types of paroxysmal dyskinesia based on their triggering events [3-4] (table 1). The prevalence of PKD is unknown, but is estimated to be present in 1/150.000 people. PKD is the most common type of the paroxysmal dyskinesias and possible overlap between categories has been described [5].

PKD can be sporadic, familial or secondary. Central nerve abnormalities, cerebral vascular insufficiency, trauma and metabolic disorders can cause symptomatic paroxysmal dyskinesia [6]. In approximately 50 percent of the cases patients have a family history [7]. The inheritance pattern is autosomal dominant and an incomplete penetrance is described (80-90%).

## **Study objective**

The purpose of this research project is to assess whether affected PKD patients have brain abnormalities compared to healthy normal controls.

Hypothesis:

Paroxysmal kinesigenic dyskinesia affected individuals exhibit abnormal spontaneous activity of the basal ganglia, thalamus and the sensorimotor cortex. Such abnormal brain activity may manifest itself as increased grey matter volume, increased white matter integrity connectivity, increased resting state activity and hyperperfusion of affected structures.

## **Study design**

Case-control study.

## **Study burden and risks**

With the screening for contraindications, no objective risks are inherent to the use of the MRI.

## **Contacts**

### **Public**

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## **Trial sites**

### **Listed location countries**

Netherlands

## **Eligibility criteria**

### **Age**

Adolescents (12-15 years)

Adolescents (16-17 years)

Adults (18-64 years)

Elderly (65 years and older)

### **Inclusion criteria**

- Diagnosis of PKD by a neurologist and clinical geneticist according the clinical criteria of Bruno et al (2004);
- Age: 16 years and older.
- Informed consent
- Able to lay still for a prolonged time (about an hour);

### **Exclusion criteria**

- Secondarily caused PKD symptoms;
- Contra-indications for MRI scanning;
- Neurocognitive disorders related to brain abnormalities or psychiatric disease.

## Study design

### Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

### Recruitment

NL	
Recruitment status:	Recruiting
Start date (anticipated):	06-09-2011
Enrollment:	40
Type:	Actual

## Ethics review

Approved WMO	
Date:	01-09-2011
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

## Study registrations

### Followed up by the following (possibly more current) registration

No registrations found.

## Other (possibly less up-to-date) registrations in this register

No registrations found.

## In other registers

### Register

CCMO

### ID

NL36982.078.11