

# Influence of ryanodyne receptor 1 mutations on pulmonary arterial pressure and ventilation during isocapnic hypoxia

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We want to investigate the influence of a mutation in the MH allele on the HPV and the normal reaction of MH patients to hypoxia

<b>Ethical review</b>	Approved WMO
<b>Status</b>	Will not start
<b>Health condition type</b>	Vascular hypertensive disorders
<b>Study type</b>	Interventional

## Summary

### ID

NL-OMON36376

### Source

ToetsingOnline

### Brief title

Ryan op pad

### Condition

- Vascular hypertensive disorders

### Synonym

pulmonary hypertension

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Leids Universitair Medisch Centrum

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

## Intervention

**Keyword:** HPV, HVR, hypoxia, ryanodine receptor

## Outcome measures

### Primary outcome

HPV

### Secondary outcome

cardiac parameters

hypoxic ventilatory response

## Study description

### Background summary

During hypoxia, a phenomenon called hypoxic pulmonary vasoconstriction, occurs. This reaction protects the body by lowering bloodflow to less ventilated parts of the lung. This protecting mechanism can also harm the patient. This is the case with COPD and the development of pulmonary hypertension. The ryanodine receptor plays an important role in HPV. MH patients have a mutation in the ryanodine receptor gene and studies in mice show a different reaction to hypoxia.

### Study objective

We want to investigate the influence of a mutation in the MH allele on the HPV and the normal reaction of MH patients to hypoxia

### Study design

case controlled intervention study

### Intervention

hypoxia

### Study burden and risks

headache during hypoxia sometimes occurs, which is treated with paracetamol  
a bruise may occur on the spot of the iv

## Contacts

### Public

Leids Universitair Medisch Centrum

albinusdreef 2  
2333 ZA, Leiden  
NL

### Scientific

Leids Universitair Medisch Centrum

albinusdreef 2  
2333 ZA, Leiden  
NL

## Trial sites

### Listed location countries

Netherlands

## Eligibility criteria

### Age

Adults (18-64 years)  
Elderly (65 years and older)

### Inclusion criteria

Healthy Malignant Hyperthermia patients with a phenotypic high susceptibility and a proven causative mutation.

The major inclusion criteria are that the subject has echo evidence of tricuspid regurgitation during systole, which is not clinically relevant but in fact can be demonstrated in most normal individuals.

## Exclusion criteria

- Obesity (BMI > 30)
- Presence of medical disease: heart-, lung-, liver-, kidney- and lung disease; diabetes
- Presence of psychiatric disease
- History of chronic alcohol or drug use
- Possibility of pregnancy
- Lactation

## Study design

### Design

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

### Recruitment

NL	
Recruitment status:	Will not start
Enrollment:	24
Type:	Anticipated

## Ethics review

Approved WMO	
Date:	23-05-2011
Application type:	First submission
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)

## 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	ID
CCMO	NL35083.058.11