

# Detection of novel genetic variants within the aquaporins 1 and 5

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| <b>Submission date</b><br>06/05/2009   | <b>Recruitment status</b><br>No longer recruiting | <input type="checkbox"/> Prospectively registered    |
|  |   | <input type="checkbox"/> Protocol                    |
| <b>Registration date</b><br>03/07/2009 | <b>Overall study status</b><br>Completed          | <input type="checkbox"/> Statistical analysis plan   |
|  |   | <input checked="" type="checkbox"/> Results          |
| <b>Last Edited</b><br>03/07/2009       | <b>Condition category</b><br>Respiratory          | <input type="checkbox"/> Individual participant data |

**Plain English summary of protocol**  
Not provided at time of registration

## Contact information

**Type(s)**  
Scientific

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## Additional identifiers

## Study information

**Scientific Title**  
Detection of novel genetic polymorphisms (single nucleotide polymorphisms [SNPs]) within the aquaporins 1 and 5: an observational single-centre study

**Study objectives**  
We hypothesised that single nucleotide polymorphisms (SNPs) within the aquaporins 1 and 5 contribute to the phenotypic variability of acute respiratory distress syndrome (ARDS), sepsis or bronchial asthma.

## **Ethics approval required**

Old ethics approval format

## **Ethics approval(s)**

Local ethics committee (Vorsitzender der Ethikkommission, Institut für Pharmakologie, Universitätsklinikum Essen) approved on the 9th December 2002 (ref: 01-97-1697, 05-2776, 06-3078, 07-3313)

## **Primary study design**

Observational

## **Study design**

Observational single-centre case-control study

## **Study type(s)**

Diagnostic

## **Health condition(s) or problem(s) studied**

Aquaporines, cell migration and inflammation

## **Interventions**

The following analyses will be performed on each participant:

1. Deoxyribonucleic acid (DNA) extraction from rest material of the routine diagnostic, e.g., saliva, urine or blood
2. Identification of novel DNA polymorphisms through sequencing polymerase chain reaction (PCR) products of the AQP1 and 5 promoter. The method of "slowdown PCR" should be used to amplify promoter fragments with extremely high GC content (greater than 85%).
3. Determination of transcriptional activity of haplotypes by reporter assays in different cell lines
4. Haplotype-dependent analysis of radiation and chemotherapeutics on cell proliferation in cell systems
5. Haplotype-dependent analysis of messenger ribonucleic acid (mRNA) level by quantitative real time PCR
6. Haplotype-dependent analysis of protein level by Western Blot
7. Haplotype-dependent analysis of cell migration
8. Haplotype-dependent analysis of transcription factors which bind to the polymorphic regions by electrophoretic mobility shift assay (EMSA)
9. Genotyping of ARDS, sepsis, bronchial asthma patients and healthy caucasian subjects by restriction fragment length polymorphism (RFLP) and pyrosequencing and haplotype-dependent analysis of survival and disease course using SPSS and GraphPad Prism software

## **Intervention Type**

Other

## **Phase**

Not Applicable

## **Primary outcome(s)**

Detection of SNPs within the Aquaporin 1 and 5 gene which contribute to the phenotypic variability of ARDS, sepsis and bronchial asthma, measured at 30 day survival and 100 day survival

## **Key secondary outcome(s)**

Analysis of survival, disease course and rehabilitation, measured at 30 day survival and 100 day survival

**Completion date**

01/01/2011

## Eligibility

**Key inclusion criteria**

1. Acute respiratory distress syndrome:
  - 1.1. Timing: acute onset
  - 1.2. Oxygenation: partial pressure of oxygen in arterial blood (PaO<sub>2</sub>)/fraction of inspired oxygen (FiO<sub>2</sub>) ratio less than 200 mmHg (regardless of positive end expiratory pressure [PEEP])
  - 1.3. Chest radiograph: bilateral infiltrates seen on frontal chest radiograph
  - 1.4. Pulmonary artery wedge (PAW): less than 18 mmHg when measured or no clinical evidence of left atrial hypertension
2. Lung function testing with body plethysmography-revealed bronchial asthma
3. Patients with severe sepsis
4. Aged 18 to 70 years, both genders

**Participant type(s)**

Patient

**Healthy volunteers allowed**

No

**Age group**

Adult

**Lower age limit**

18 Years

**Sex**

All

**Key exclusion criteria**

No written informed consent is obtained

**Date of first enrolment**

01/01/2005

**Date of final enrolment**

01/01/2011

## Locations

**Countries of recruitment**

Germany

**Study participating centre**  
Klinik für Anesthesiologie und Intensivmedizin  
Essen  
Germany  
45122

## Sponsor information

### Organisation

German Research Council (Deutsche Forschungsgemeinschaft [DFG]) (Germany)

### ROR

<https://ror.org/018mejw64>

## Funder(s)

### Funder type

Research council

### Funder Name

German Research Council (Deutsche Forschungsgemeinschaft [DFG]) (Germany) - pending as of 06/05/2009

## Results and Publications

### Individual participant data (IPD) sharing plan

### IPD sharing plan summary

Not provided at time of registration

### Study outputs

| Output type                     | Details | Date created | Date added | Peer reviewed? | Patient-facing? |
|---------------------------------|---------|--------------|------------|----------------|-----------------|
| <a href="#">Results article</a> | results | 01/11/2008   |            | Yes            | No              |