Trial Description

Title

Genotype-PhenotypeRelationsship in Sarcoidosis

Trial Acronym

GenoPhenoReSa

URL of the trial

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Brief Summary in Lay Language

The influence of the genetic background on the clinical phenotype will be investigated.

Brief Summary in Scientific Language

Sarcoidosis is a systemic granulomatous disorder of unknown aetiology, preferentially affecting the lung. The disease has a wide spectrum of clinical courses, ranging from spontaneous remission to disabling organ damage or even death. There is accumulating evidence for a genetic susceptibility to sarcoidosis, including an association with genes in the major histocompatibility complex (MHC). In a genome-wide linkage study, we recognized seven chromosomal regions of aetiological relevance and recently identified BTNL2 as a new sarcoidosis disease gene on chromosome 6. In a genome-wide association study using 100K Affymetrix micro-arrays, we uncovered two additional susceptibility regions on chromosomes 7 and 10. In the present project, 2000 sarcoidosis patients will be extracted from a cohort of 5570 prevalent cases and genotyped for MHC genes, BTNL2, and an estimated 15 new positional candidate genes from our genome-wide 500K SNP chip studies and ongoing replication. Patients will be phenotyped at 31 study sites according to a standardized protocol and the relationship between their phenotype and genotype will be assessed. In particular, genotypes of patients with rare, unfavourable and chronic disease courses, including cardiac, neurological, cutaneous, and therapy-resistant manifestations, will be compared to those associated with spontaneous resolution. The patient cohort will be large enough to contain a suffi-ciently large number of rare phenotypes so as to ensure prognostic usefulness of the respective results. In practise, patients with adverse genotypes should be intensely monitored and would benefit most from new therapeutic approaches.

Do you plan to share individual participant data with other researchers?

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Description IPD sharing plan

[---]*

Organizational Data

- DRKS-ID: **DRKS00000045**
- Date of Registration in DRKS: **2008/11/18**
- Date of Registration in Partner Registry or other Primary Registry: **2006/09/19**
- Investigator Sponsored/Initiated Trial (IST/IIT): **yes**
- Ethics Approval/Approval of the Ethics Committee: **Approved**
- (leading) Ethics Committee Nr.: **005/06 , Ethik-Kommission der Albert-Ludwigs-Universität Freiburg**

Secondary IDs

- Partner Registry-ID: **UKF000874 (Register Klinischer Studien des Universitätsklinikums Freiburg)**

Health condition or Problem studied

- ICD10: **D86.0 - Sarcoidosis of lung**

Interventions/Observational Groups

- Arm 1: **taking of a blood sample, genotypification and phänotypification**

Characteristics

- Study Type: **Non-interventional**
- Study Type Non-Interventional: **Observational study**
- Allocation: **[---]***
- Blinding: **[---]***
- Who is blinded: **[---]***
- Control: **Uncontrolled/Single arm**
- Purpose: **Basic research/physiological study**
- Assignment: **[---]***
- Phase: **[---]***
- Off-label use (Zulassungsüberschreitende Anwendung eines Arzneimittels): **[---]***

Primary Outcome

Relationship between candidate genes and clinical outcome

Secondary Outcome

[---]*

Countries of recruitment

- DE **Germany**
- IS **Iceland**
- DK **Denmark**
- RU **Russian Federation**
- IE **Ireland**
- UK **United Kingdom**
- BE **Belgium**
- NL **Netherlands**
- PL **Poland**
- FR **France**
- CH **Switzerland**
- CZ **Czech Republic**
- HU **Hungary**
- SI **Slovenia**
- RS **Serbia**
- IT **Italy**

Locations of Recruitment

Recruitment

- Planned/Actual: **Actual**
- (Anticipated or Actual) Date of First Enrollment: **2008/10/13**
- Target Sample Size: **2000**
- Monocenter/Multicenter trial: **Multicenter trial**
-

Planned/Actual: **Actual**

(Anticipated or Actual) Date of First Enrollment: **2008/10/13**

Target Sample Size: **2000**

Monocenter/Multicenter trial: **Multicenter trial**

National/International: **International**

Inclusion Criteria

- Gender: **Both, male and female**
- Minimum Age: **no minimum age**
- Maximum Age: **no maximum age**

Additional Inclusion Criteria

Sarcoidosis of any clinical phenotype diagnosed according to ATS/ERS criteria

Exclusion criteria

No sarcoidosis

Addresses

- **Primary Sponsor**

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Sources of Monetary or Material Support

■ **Public funding institutions financed by tax money/Government funding body (German Research Foundation (DFG), Federal Ministry of Education and Research (BMBF), etc.)**

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URL: **www.dfg.de**

Status

■ Recruitment Status: **Recruiting complete, follow-up complete**

■

Study Closing (LPLV): **2012/03/30**

Trial Publications, Results and other documents

** This entry means the parameter is not applicable or has not been set.*

**** This entry means that data is not displayed due to insufficient data privacy clearing.*