Trial Description

Title

Epidemiology, diagnosis and clinical picture of PHACE Syndrome and Sturge-Weber Syndrome in Germany, Switzerland and Austria

Trial Acronym

[---]*

URL of the trial

http://www.uks.eu/fileadmin/UKS/Einrichtungen/Kliniken_und_Institute/Kinder_und_Jugenmedizin/Kinderklinik/Forschung/201711-ESNEK_Studienexpose_kurz_final.pdf

Brief Summary in Lay Language

This study aims to collect data on the frequency and the distribution of the two diseases “Sturge-Weber Syndrome” and “PHACE Syndrome” in the German-speaking population. These two rare neurologic diseases in children are so-called neurocutaneous diseases, i.e. they primarily affect the skin and the brains. Potential study participants are all children up to the age of 18 years with either of the two diagnoses and place of residence in Germany, Switzerland or Austria. We use a mailing list which includes all pediatricians who are specialists for neurologic diseases in children (neuropediatricians) to identify potential study participants. This mailing list is a research instrument developed by pediatricians from University Medical Center Göttingen and it includes all neuropediatricians from Germany, Switzerland and Austria. Via E-Mail, all neuropediatricians are asked to anonymously report how many patients they have treated with either of the two diagnoses in the past years. If the patients agree to participate in the study, we contact them and we assess data for the study with the help of a questionnaire. The questionnaire was developed in cooperation with the PHACE Registry at Children's Hospital of Wisconsin, USA.
One of study objectives is to calculate the proportion of disease within the investigated population (i.e. the prevalence) of these two rare, probably genetic diseases. Additionally, we investigate which anomalies are known before birth and we study the current state of diagnostic procedures and genetic testing. We also collect data on clinical symptoms and current therapies.

Brief Summary in Scientific Language

The main objective of the study is to collect data on the epidemiology of the phacomatoses Sturge-Weber Syndrome and PHACE Syndrome and estimate their prevalences in the German-speaking countries (Germany, Switzerland, Austria). We also collect data on prenatal anomalies and the current state of diagnostic procedures including genetic analyses. Additionally, we assess data on the clinical picture and therapeutic measures.
The study is performed in cooperation with the “ESNEK” Network (German: Erhebung Seltener Neurologischer Erkrankungen im Kindesalter; English: Assessment Of Rare Neurologic Diseases In Childhood), located at University Medical Center Göttingen: Via a mailing list, all neuropediatricians in Germany,

Switzerland and Austria are asked to identify potential study participants. Medical data are assessed using a questionnaire. We developed the questionnaire in cooperation with the PHACE registry at Children's Hospital of Wisconsin, USA. The study was approved by the ethics committee Saarbrücken.

Organizational Data

- DRKS-ID: **DRKS00013551**
- Date of Registration in DRKS: **2017/12/27**
- Date of Registration in Partner Registry or other Primary Registry: **[---]***
- Investigator Sponsored/Initiated Trial (IST/IIT): **yes**
- Ethics Approval/Approval of the Ethics Committee: **Approved**
- (leading) Ethics Committee Nr.: **209/17 , Ethik-Kommission bei der Ärztekammer des Saarlandes**

Secondary IDs

- Universal Trial Number (UTN): **U1111-1206-9923**

Health condition or Problem studied

- ICD10: **Q28.8 - Other specified congenital malformations of circulatory system**
- ICD10: **Q85.8 - Other phakomatoses, not elsewhere classified**

Interventions/Observational Groups

- Arm 1: **Parents with a child affected by PHACE Syndrome or Sturge-Weber Syndrome once receive a questionnaire on the child's symptoms, the diagnostic procedures carried out until now, anomalies known before birth, response to treatment**

Characteristics

- Study Type: **Non-interventional**
- Study Type Non-Interventional: **Epidemiological study**
- Allocation: **Single arm study**
- Blinding: **[---]***
- Who is blinded: **[---]***
- Control: **Uncontrolled/Single arm**
- Purpose: **Basic research/physiological study**

Study Type: **Non-interventional**

Study Type Non-Interventional: **Epidemiological study**

Allocation: **Single arm study**

Blinding: **[---]***

Who is blinded: **[---]***

Control: **Uncontrolled/Single arm**

Purpose: **Basic research/physiological study**

- Assignment: **Single (group)**
- Phase: **N/A**
- Off-label use (Zulassungsüberschreitende Anwendung eines Arzneimittels): **N/A**

Primary Outcome

Prevalence of PHACE Syndrome and Sturge-Weber Syndrome in Germany, Switzerland and Austria
(treatment prevalence).
As a first step, we contact neuropediatricians in Germany, Switzerland and Austria via email, who recruit affected families. Then, we assess medical data via questionnaire handed out to the families and neuropediatricians.

Secondary Outcome

Data assessment via once-only questionnaire handed out to affected families and neuropediatricians: prenatal risk factors and anomalies,
clinical symptoms, diagnostic procedures including genetic analyses, therapy strategies and their effectiveness

Countries of recruitment

- DE **Germany**
- CH **Switzerland**
- AT **Austria**

Locations of Recruitment

- other **Alle Kliniken und Praxen, in denen Kinderneurologen tätig sind / All hospitals and medical practices in which neuropediatricians work, Deutschland, Schweiz, Österreich / Germany, Switzerland, Austria**

Recruitment

- Planned/Actual: **Actual**
- (Anticipated or Actual) Date of First Enrollment: **2018/01/25**
- Target Sample Size: **150**
- Monocenter/Multicenter trial: **Multicenter trial**
- National/International: **International**

Inclusion Criteria

- Gender: **Both, male and female**
- Minimum Age: **no minimum age**
- Maximum Age: **18 Years**

Additional Inclusion Criteria

Positive Diagnosis of "PHACE Syndrome" OR "Sturge-Weber Syndrome" by a medical doctor, e.g. pediatrician, family physician, geneticist

Exclusion criteria

None

Addresses

- **Primary Sponsor**

  Klinik für Allgemeine Pädiatrie und Neonatologie, Universitätsklinikum des Saarlandes
  66421 Homburg/Saar
  Germany

  Telephone: **[---]***
  Fax: **[---]***
  E-mail: **[---]***
  URL: **http://www.uks.eu/de/einrichtungen/kliniken_institute/kinder-und-jugendmedizin/klinik-fuer-allgemeine-paediatrie-und-neonatologie**
- **Contact for Scientific Queries**

  Klinik für Allgemeine Pädiatrie und NeonatologieUniversitätsklinikum des Saarlandes
  Ms. Dr.med. Sigrid Disse
  Kirrberger Straße 100
  66421 Homburg/ Saar
  Germany

  Telephone: **+49 6841 1628000**

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Ms. Dr.med. Sigrid Disse
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Germany

Telephone: **+49 6841 1628000**
Fax: **[---]***
E-mail: **sigrid.disse at uks.eu**
URL: **http://www.uks.eu/de/einrichtungen/kliniken_institute/kinder-und-jugendmedizin/klinik-fuer-allgemeine-paediatrie-und-neonatologie**

■ **Contact for Public Queries**

Klinik für Allgemeine Pädiatrie und NeonatologieUniversitätsklinikum des Saarlandes
Ms. Dr.med. Sigrid Disse
Kirrberger Straße 100
66421 Homburg/ Saar
Germany

Telephone: **+49 6841 1628000**
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■ **Collaborator, Other Address**

Zentrum für Seltene Neurologische Erkrankungen im Kindesalter (GoRare), Sozialpädiatrisches Zentrum, Klinik für Kinder- und Jugendmedizin, Universitätsmedizin Göttingen
Robert-Koch-Straße 40
37075 Göttingen
Germany

Telephone: **[---]***
Fax: **[---]***
E-mail: **[---]***
URL: **[---]***

Sources of Monetary or Material Support

■ **Institutional budget, no external funding (budget of sponsor/PI)**

Klinik für Allgemeine Pädiatrie und NeonatologieUniversitätsklinikum des Saarlandes
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66421 Homburg/ Saar

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Status

- Recruitment Status: **Recruiting ongoing**
- Study Closing (LPLV): **[---]***

Trial Publications, Results and other documents

Please note:
There are additional attributes available concerning this trial. To open an extended view please click here.