PLEASE NOTE: *This study has been imported from ClinicalTrials.gov without additional data checks.*

Trial Description

Title

Pyruvate Kinase Deficiency (PKD) Natural History Study

Trial Acronym

PKD NHS

URL of the trial

[---]*

Brief Summary in Lay Language

The purpose of this study is to describe the range and incidence of symptoms, treatments, and complications related to pyruvate kinase deficiency (PKD). Eligible patients are those of all ages with known PKD or with a hemolytic anemia and a family member with PKD. The study will collect retrospective medical history, routine clinical care data, and quality of life measures at baseline and annually for patients with PKD.

Brief Summary in Scientific Language

The purpose of the Pyruvate Kinase Deficiency (PKD) Natural History Study is to describe the natural history of PKD and the range and incidence of symptoms, treatments, and complications related to PKD. The study will collect retrospective medical history and routine clinical care data at baseline and annually for patients with PKD. Patients without a genetic diagnosis will have a blood sample drawn for genetic diagnostic confirmation for research purposes. Understanding the clinical variation among participants with PKD, and assessing treatments specific to PKD and their outcomes will accelerate improvement in the care of patients with PKD. Understanding the natural history of PKD may be useful in the design of future interventional studies. Detailed genotypic and phenotypic characterization of the cohort will allow for continued in depth characterization of PKD. Finally,

the PKD Natural History Study will identify interested participants for future PKD studies.

Primary Objectives:

1. To estimate the transfusion burden in splenectomized and non-splenectomized participants with PKD.

2. To establish a patient registry as a potential source for recruitment to future research studies in PKD.

Secondary Objectives:

1. To determine if patient-reported outcomes, including quality of life and fatigue scales, are associated with age, genotype, hemoglobin nadir, and/or transfusion burden, overall and within the subgroups of splenectomized vs. non-splenectomized participants;

2. To describe changes over time in the range of hemoglobin values and markers of hemolysis within individual participants and among participants with PKD;

3. To estimate the incidence of past splenectomy and annual splenectomy rate, as treatment for PKD;

4. To estimate the prevalence and severity and describe the treatment of hepatic and cardiac iron overload and its complications in PKD (liver, cardiac, growth defects, hypogonadotropic hypogonadism, and other endocrine defects). To describe the changes in these complications that may occur over time and by age group;

5. To estimate the prevalence of co-morbidities associated with chronic hemolysis in PKD, to identify which co-morbidities are the most common, and to determine if the prevalence and/or severity of co-morbidities change over time and by age at the time of the first appearance of the co-morbidity;

6. To determine pregnancy outcomes among participants with PKD;

7. To describe genotypic and phenotypic variation among participants and explore genotype-phenotype correlation in PKD.

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

[---]*

Description IPD sharing plan

[---]*

Organizational Data

- DRKS-ID: **DRKS00006537**
- Date of Registration in DRKS: **2014/07/23**
- Date of Registration in Partner Registry or other Primary Registry: **2014/01/28**
- Investigator Sponsored/Initiated Trial (IST/IIT): **yes**
- Ethics Approval/Approval of the Ethics Committee: **[---]***
- (leading) Ethics Committee Nr.: **[---]***

Secondary IDs

- Primary Registry-ID: **NCT02053480 (ClinicalTrials.gov)**
- Sponsor-ID: **P00010515 (Children's Hospital Boston)**

Health condition or Problem studied

- Free text: **Pyruvate Kinase Deficiency**
- Free text: **Congenital Non-Spherocytic Hemolytic Anemia**
- ICD10: **D55.2 - Anaemia due to disorders of glycolytic enzymes**

Interventions/Observational Groups

Characteristics

- Study Type: **Non-interventional**
- Study Type Non-Interventional: **Observational study**
- Allocation: **[---]***
- Blinding: **[---]***
- Who is blinded: **[---]***
- Control: **[---]***
- Purpose: **[---]***
-

Study Type: **Non-interventional**

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

Allocation: **[---]***

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

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

Control: **[---]***

Purpose: **[---]***

Assignment: **[---]***

- Phase: **N/A**
- Off-label use (Zulassungsüberschreitende Anwendung eines Arzneimittels): **[---]***

Primary Outcome

- transfusion burden in splenectomized and non-splenectomized participants; time frame: 12 weeks

Secondary Outcome

- patient-reported outcomes; time frame: enrollment, annually, up to 2 years; EuroQoL-5D-5L, Functional Assessment of Cancer Therapy-Anemia (FACT-An), Pediatric Quality of Life Inventory 4.0 (pedsQL 4.0), Pediatric Functional Assessment of Chronic Illness-Fatigue (pedsFACIT-F), Patient Reported Outcomes Measurement Information System Fatigue (PROMIS Fatigue)
- changes over time in hemoglobin and markers of hemolysis; time frame: enrollment, annually, up to 2 years
- prevalence and severity of iron overload; time frame: enrollment, annually, up to 2 years

Countries of recruitment

- US **United States**

Locations of Recruitment

Recruitment

- Planned/Actual: **[---]***
- (Anticipated or Actual) Date of First Enrollment: **2013/12/31**
- Target Sample Size: **100**
- 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

- Patients of all ages with biochemically or genetically diagnosed PKD.

- Patients with a hemolytic anemia AND a family member with genetically diagnosed PKD

- The participant or the guardian of the participant is willing and able to give written informed consent and/or assent.

Exclusion criteria

- The participant or the guardian of the participant is unwilling or unable to give written informed consent and/or assent.

Addresses

- **Primary Sponsor**

  Children's Hospital Boston

  Telephone: **[---]***
  Fax: **[---]***
  E-mail: **[---]***
  URL: **[---]***
- **Contact for Scientific Queries**

  Rachael F Grace, MD, MMSc

Contact for Scientific Queries

Rachael F Grace, MD, MMSc

Telephone: **617-632-3528**
Fax: **[---]***
E-mail: **Rachael.Grace at childrens.harvard.edu**
URL: **[---]***

- **Contact for Public Queries**

  Rachael F Grace, MD, MMSc

  Telephone: **617-632-3528**
  Fax: **[---]***
  E-mail: **Rachael.Grace at childrens.harvard.edu**
  URL: **[---]***

- **Collaborator, Other Address**

  Agios Pharmaceuticals, Inc.

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

Sources of Monetary or Material Support

- **[---]***

  Bitte wenden Sie sich an den Sponsor / Please refer to primary sponsor

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

Status

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

Trial Publications, Results and other documents

The parameters in ClinicalTrials.gov and DRKS are not identical. Therefore the data import from ClinicalTrials.gov required adjustments. For full details please see the DRKS FAQs.

- Translation on version: 3

- Last processed date by ClinicalTrials.gov: 2014/07/20

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