

Pyruvate Kinase Deficiency (PKD): Survey on facilities available for diagnosis

***Data that will be published in ERN-EuroBloodNet website under your authorization**

A. Organization Data

Institution*:

Website*:

Department*:

Department e-mail*:

Department phone*:

Contact person*:

Contact person e-mail:

Contact person phone:

Are you a clinician (hematologist, pediatrician)?: Yes/No

Are you a diagnostician (medical doctor, biologist, researcher)?: Yes/No

B. PKD Patients

1. Number of patients affected by PKD currently followed in your centre*:

2. Number of patients affected by PKD currently followed in your centre with genetic diagnosis*:

3. Mean number of new patients per year affected by PKD followed in your centre*:

4. Does your centre participate in a database/registry for PK deficiency?: Yes/No

- If yes:

a. Database/Registry title:

b. If available, link to Database/Registry website:

c. Type of registry:

- i. National Registry
- ii. International Registry
- iii. Regional Registry
- iv. Hospital / Laboratory database
- v. Other: (Specify)

d. Curator Name:

e. Curator e-mail:

C. PKD Diagnosis - Part A PK enzyme activity

1. Total number of PKD diagnosis*:

2. Mean number of new PKD diagnosis per year*:

3. Do you perform in your centre PK activity assay?*: Yes/No

- If yes:

a. Which method do you use to perform PK activity assay:*

- i. Spectrophotometric assay (Beuter 1980)
- ii. Other quantitative methods (please specify):
- iii. Semi-quantitative methods (please specify):
- iv. Commercial kit (please specify):
- v. Other (please specify):

b. Do you also perform hexokinase (HK) enzyme activity to assess mean red cell age through PK/HK ratio?: Yes/No

4. Do you externalize the PK activity quantitative assay?*: Yes/No

- If yes:

a. External Institution/Laboratory Name (Not mandatory):

b. Which method is used to perform PK activity assay: Multiple choice

- i. Spectrophotometric assay (Beuter 1980)
- ii. Other quantitative methods (please specify):
- iii. Semi-quantitative methods (please specify):
- iv. Commercial kit (please specify):
- v. Other (please specify):
- vi. I am not informed

b. Do you also externalize hexokinase (HK) enzyme activity to assess mean red cell age through PK/HK ratio? Yes/No

5. If you do not perform PK activity assay nor in your centre neither externalized please specify the reason:

- a. We only perform DNA analysis of PKLR
- b. PK activity test is not authorized in my centre to be externalized
- c. PK activity assay is not available in my country (to the best of my knowledge)
- d. Other (Please, specify):

D. PKD Diagnosis - Part B PKLR genetic analysis

1. Do you always confirm a decreased PK activity at molecular level?: Yes/No

If not always, please specify in which cases:

2. Do you perform in your centre PKLR genetic characterization?*: Yes/No

If yes:

- a. Which method is used for PKLR genetic characterization*
 - i. Sanger
 - ii. NGS
 - iii. Other (please specify):

3. Do you externalize the PKLR genetic characterization?*: Yes/No

If yes:

- a. External Institution/Laboratory Name (Not mandatory):
- b. Which method is used for PKLR genetic characterization:
 - iv. Sanger
 - v. NGS
 - vi. Other (please specify):
 - vii. I am not informed

E. Genetic counselling and prenatal diagnosis

1. Genetic counselling for PKD is offered in your centre*?: Yes/No

2. Is prenatal diagnosis for PKD available in your centre*? Yes/No

Do you give your consent to publish data with* in the ERN-EuroBloodNet website

www.eurobloodnet.eu ? Yes/No