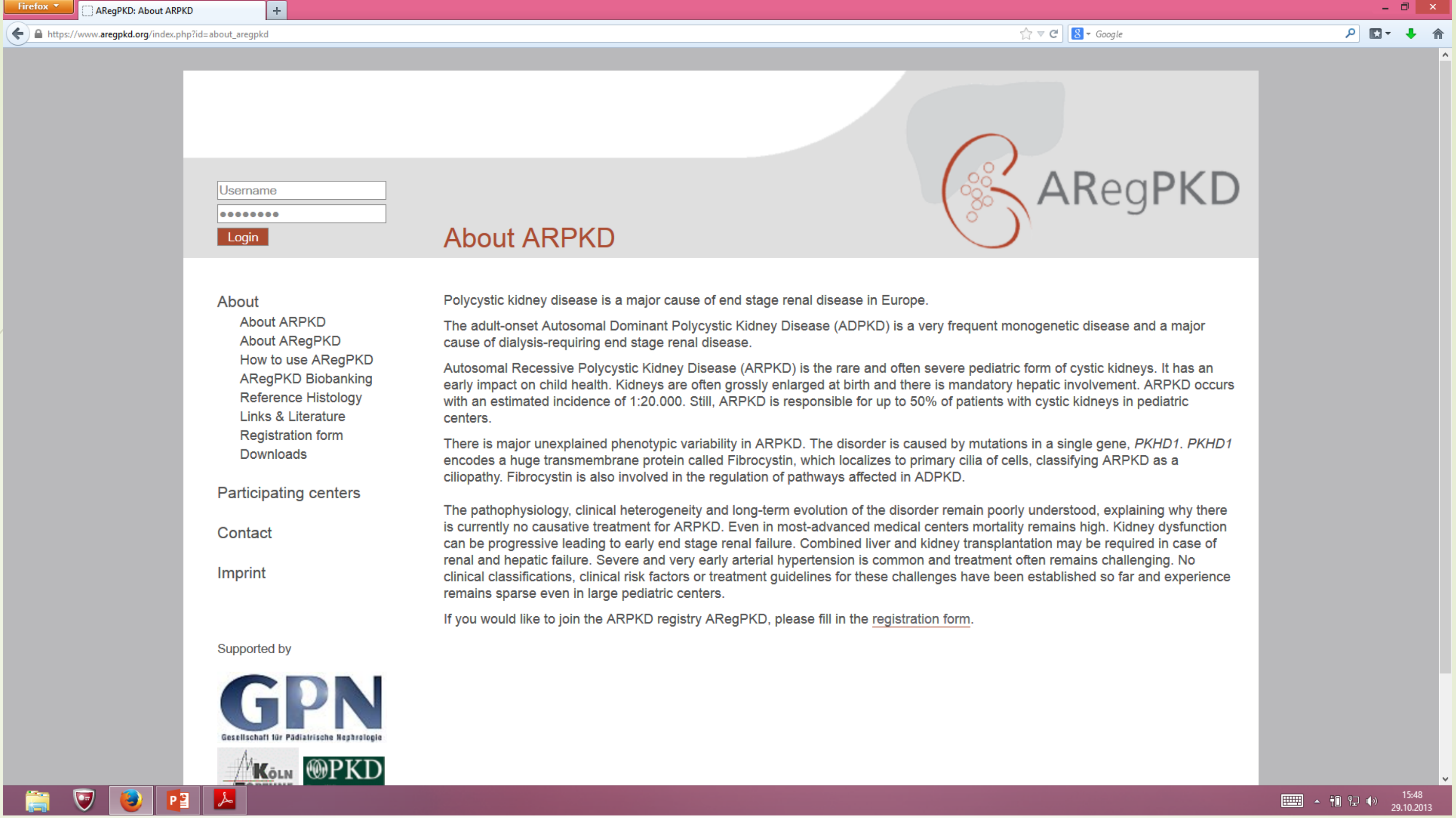
ARegPKD

A small handbook for ARegPKD



About ARPKD

Polycystic kidney disease is a major cause of end stage renal disease in Europe.

The adult-onset Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a very frequent monogenetic disease and a major cause of dialysis-requiring end stage renal disease.

Autosomal Recessive Polycystic Kidney Disease (ARPKD) is the rare and often severe pediatric form of cystic kidneys. It has an early impact on child health. Kidneys are often grossly enlarged at birth and there is mandatory hepatic involvement. ARPKD occurs with an estimated incidence of 1:20.000. Still, ARPKD is responsible for up to 50% of patients with cystic kidneys in pediatric centers.

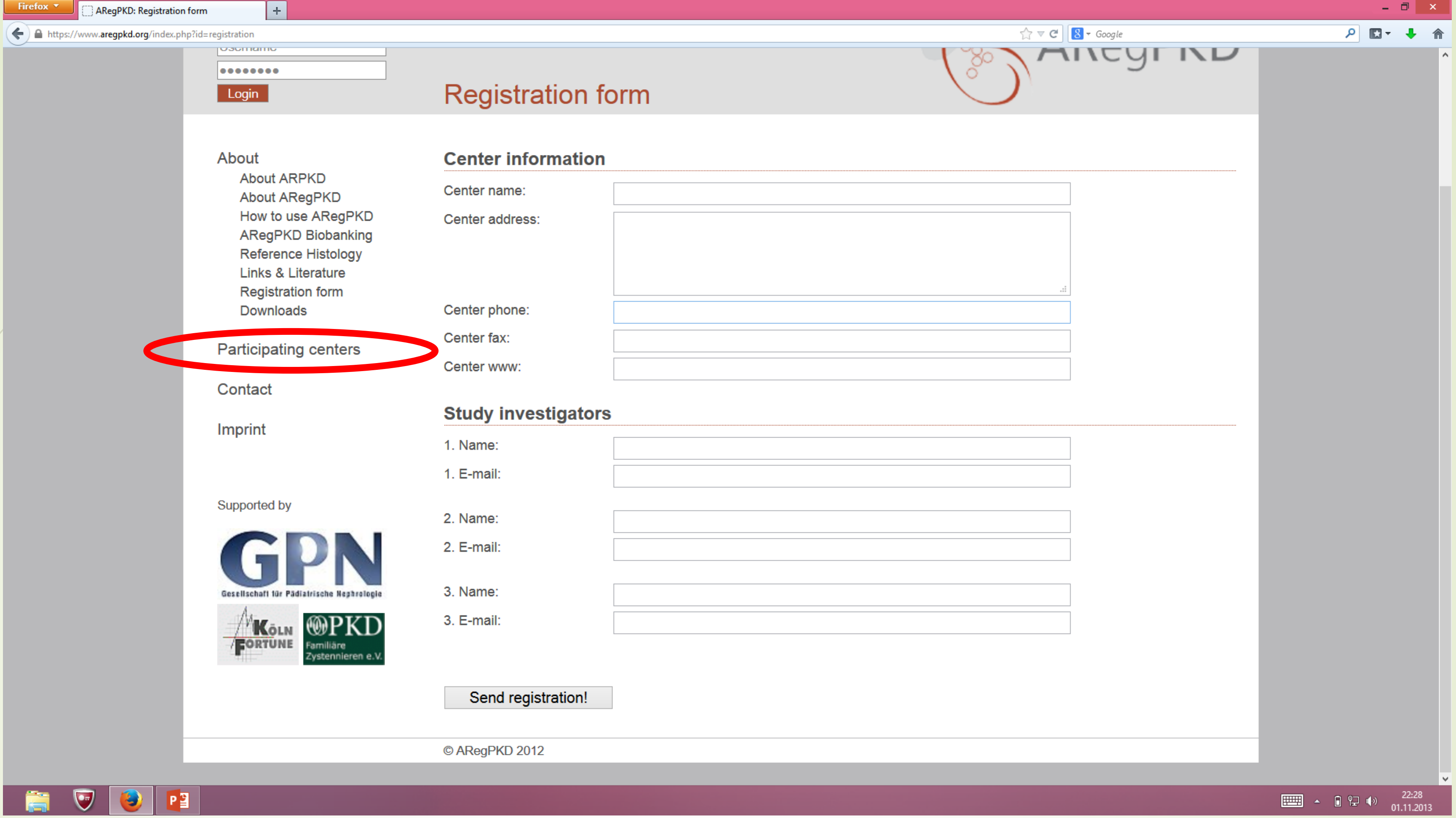
There is major unexplained phenotypic variability in ARPKD. The disorder is caused by mutations in a single gene, *PKHD1*. *PKHD1* encodes a huge transmembrane protein called Fibrocystin, which localizes to primary cilia of cells, classifying ARPKD as a ciliopathy. Fibrocystin is also involved in the regulation of pathways affected in ADPKD.

The pathophysiology, clinical heterogeneity and long-term evolution of the disorder remain poorly understood, explaining why there is currently no causative treatment for ARPKD. Even in most-advanced medical centers mortality remains high. Kidney dysfunction can be progressive leading to early end stage renal failure. Combined liver and kidney transplantation may be required in case of renal and hepatic failure. Severe and very early arterial hypertension is common and treatment often remains challenging. No clinical classifications, clinical risk factors or treatment guidelines for these challenges have been established so far and experience remains sparse even in large pediatric centers.

If you would like to join the ARPKD registry ARegPKD, please fill in the registration form.

Enter www.aregpkd.org and visit our website.

Enter Username and password - both will be sent to you after registration in an e-mail.



Registration form

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Center information

Center name:
Center address:
Center phone:
Center fax:
Center www:

Study investigators

1. Name:
1. E-mail:

2. Name:
2. E-mail:

3. Name:
3. E-mail:

Send registration!

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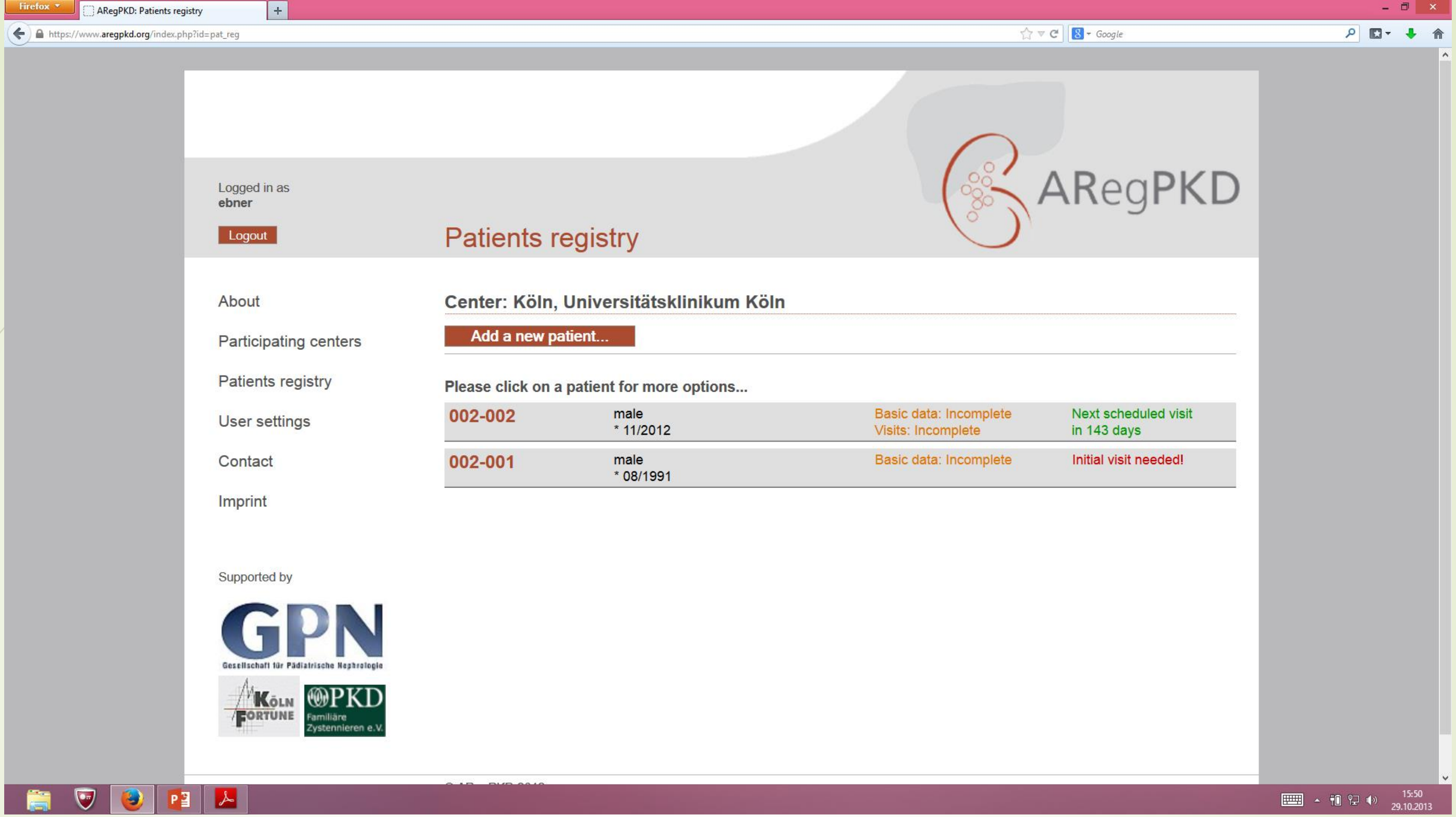
In order to register you need to visit the *Registration form* and fill in the form. We will send you a username and password as soon as possible.

Once you have registered please make sure that **a positive votum of the corresponding ethics committee** has been obtained and that your patient and/or a representative has received, understood and signed the patient information and **informed consent** form.



Logged in as
ebner

Patients registry

Center: Köln, Universitätsklinikum Köln

Add a new patient...

Please click on a patient for more options...

002-002	male * 11/2012	Basic data: Incomplete Visits: Incomplete	Next scheduled visit in 143 days
002-001	male * 08/1991	Basic data: Incomplete	Initial visit needed!

After logging in, you can either *Add a new patient* or work on registered patients.

Basic data

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Please select...	*Status*
Basic information	-EMPTY-
Family history	-EMPTY-
Pre- and perinatal period / infancy	-EMPTY-
Initial diagnosis	-EMPTY-
Genetics	-EMPTY-

Import basic data from 4C-Registry (*.dat)
Browse for the data file and press the import button.
Only possible before saving!
Keine Datei ausgewählt.

Patient-ID Will be generated after saving

IMPORTANT: Please don't forget to record the Patient-ID in your study documentation for future identification of this patient!

Basic information

Date of informed consent* (DD/MM/YYYY)

Date of birth* (MM/YYYY)

Sex*

Dates

Onset of symptoms (incl. prenatal symptoms) (DD/MM/YYYY)[1] ☐ unknown

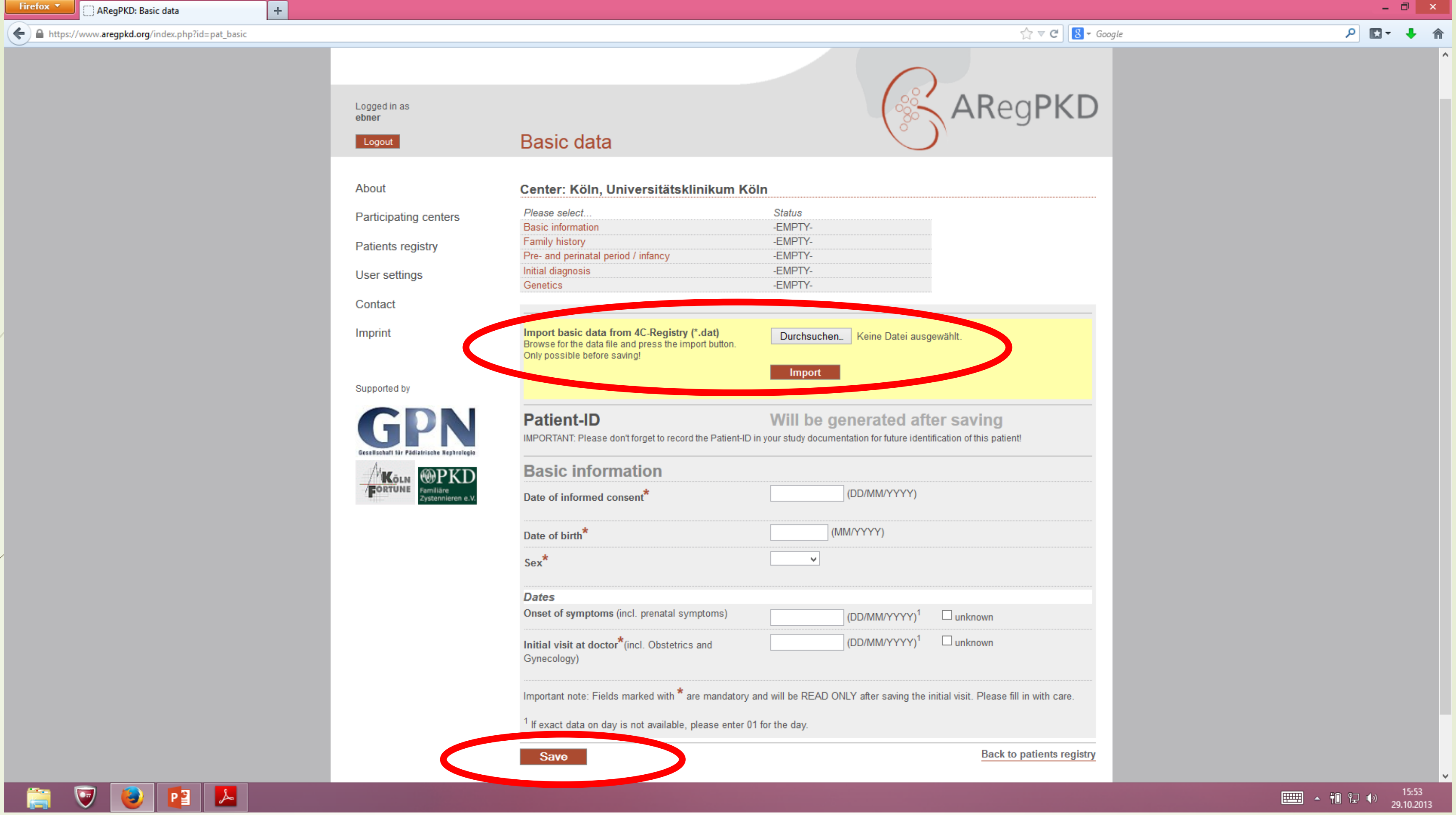
Initial visit at doctor* (incl. Obstetrics and Gynecology) (DD/MM/YYYY)[1] ☐ unknown

Important note: Fields marked with * are mandatory and will be READ ONLY after saving the initial visit. Please fill in with care.

[1] If exact data on day is not available, please enter 01 for the day.

When you *Add a new patient*, you are directed to the *Basic data* section, which you need to fill in only once. *Basic data* covers general information on your patient and on the ARPKD diagnosis.

Please note: You can only continue after entering the *Date of informed consent*! Questions marked with * are mandatory and will be READ ONLY after saving the initial visit. Please fill in with care. Mandatory questions exist in the *Basic Information* and *Initial diagnosis* subfolders.



Basic data

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Please select...	*Status*
Basic information	-EMPTY-
Family history	-EMPTY-
Pre- and perinatal period / infancy	-EMPTY-
Initial diagnosis	-EMPTY-
Genetics	-EMPTY-

Import basic data from 4C-Registry (*.dat)
Browse for the data file and press the import button.
Only possible before saving!

Durchsuchen... Keine Datei ausgewählt.

Import

Patient-ID Will be generated after saving

IMPORTANT: Please don't forget to record the Patient-ID in your study documentation for future identification of this patient!

Basic information

Date of informed consent*	(DD/MM/YYYY)
Date of birth*	(MM/YYYY)
Sex*	

Dates

Onset of symptoms (incl. prenatal symptoms)	(DD/MM/YYYY)[1]	☐ unknown
Initial visit at doctor*(incl. Obstetrics and Gynecology)	(DD/MM/YYYY)[1]	☐ unknown

Important note: Fields marked with * are mandatory and will be READ ONLY after saving the initial visit. Please fill in with care.

[1] If exact data on day is not available, please enter 01 for the day.

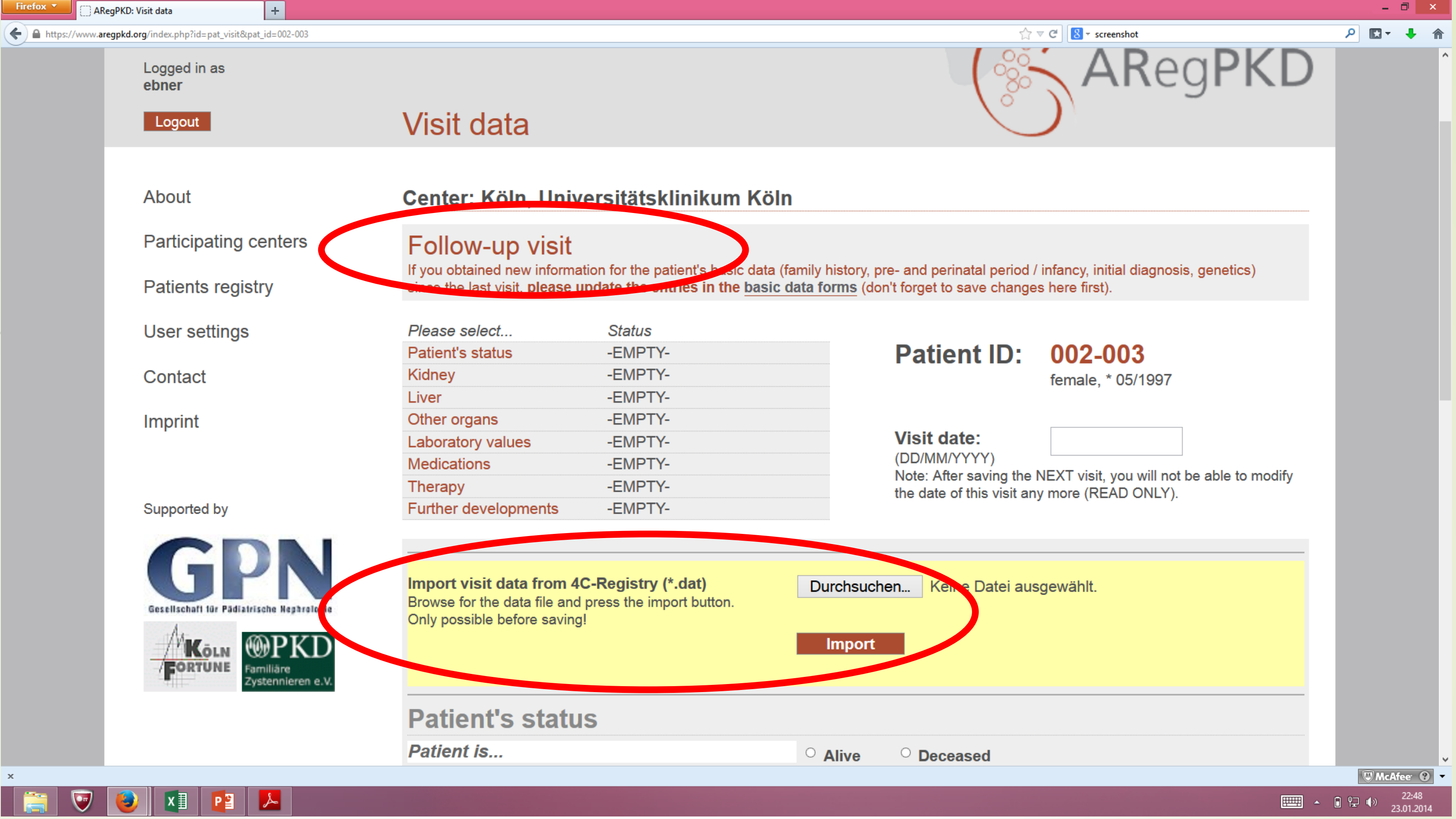
Save

Back to patients registry

For patients participating in the 4C-study: You can import data from the 4C-study. To do so please open your patient's file of the 4C-study. You will find an *Export for ARegPKD*-button for every visit (and basic data). Please save the Export file e.g. on your desktop.

You can now import the data into ARegPKD. To do so browse for the data file and press the *Import* button. This import is only possible before saving. If you imported data these will be automatically filled in matching fields.

Don`t forget to press *Save* after entering your data!



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Visit data

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Follow-up visit

If you obtained new information for the patient's basic data (family history, pre- and perinatal period / infancy, initial diagnosis, genetics) since the last visit, **please update the entries in the basic data forms** (don't forget to save changes here first).

Please select...	*Status*
Patient's status	-EMPTY-
Kidney	-EMPTY-
Liver	-EMPTY-
Other organs	-EMPTY-
Laboratory values	-EMPTY-
Medications	-EMPTY-
Therapy	-EMPTY-
Further developments	-EMPTY-

Patient ID: 002-003
female, * 05/1997

Visit date:
(DD/MM/YYYY)
Note: After saving the NEXT visit, you will not be able to modify the date of this visit any more (READ ONLY).

Import visit data from 4C-Registry (*.dat)
Browse for the data file and press the import button.
Only possible before saving!

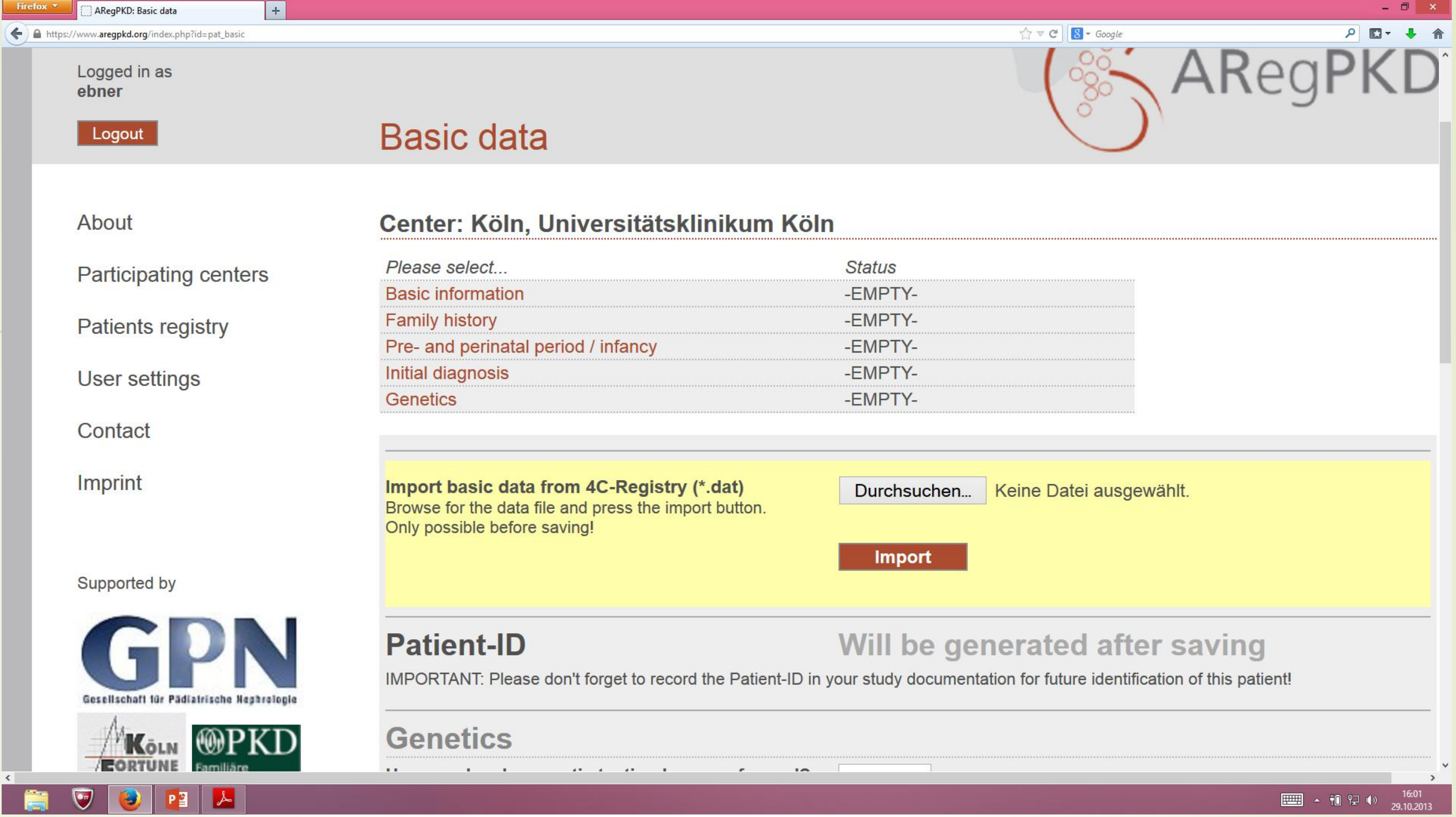
Keine Datei ausgewählt.

Patient's status

Patient is... ○ Alive ○ Deceased

Please note: it is advised to import data from the 4C-study when you start entering a patient`s data. If you want to import data for upcoming visits, this is no problem: go to *Add next follow-up visit* and you will find the yellow field with *Import* button automatically.

If you want to import data for visits that you already have entered, please write us an email as this option is available only for admininstration staff!



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Basic data

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Please select...	*Status*
Basic information	-EMPTY-
Family history	-EMPTY-
Pre- and perinatal period / infancy	-EMPTY-
Initial diagnosis	-EMPTY-
Genetics	-EMPTY-

Import basic data from 4C-Registry (*.dat)
Browse for the data file and press the import button.
Only possible before saving!

Keine Datei ausgewählt.

Patient-ID **Will be generated after saving**

IMPORTANT: Please don't forget to record the Patient-ID in your study documentation for future identification of this patient!

Genetics

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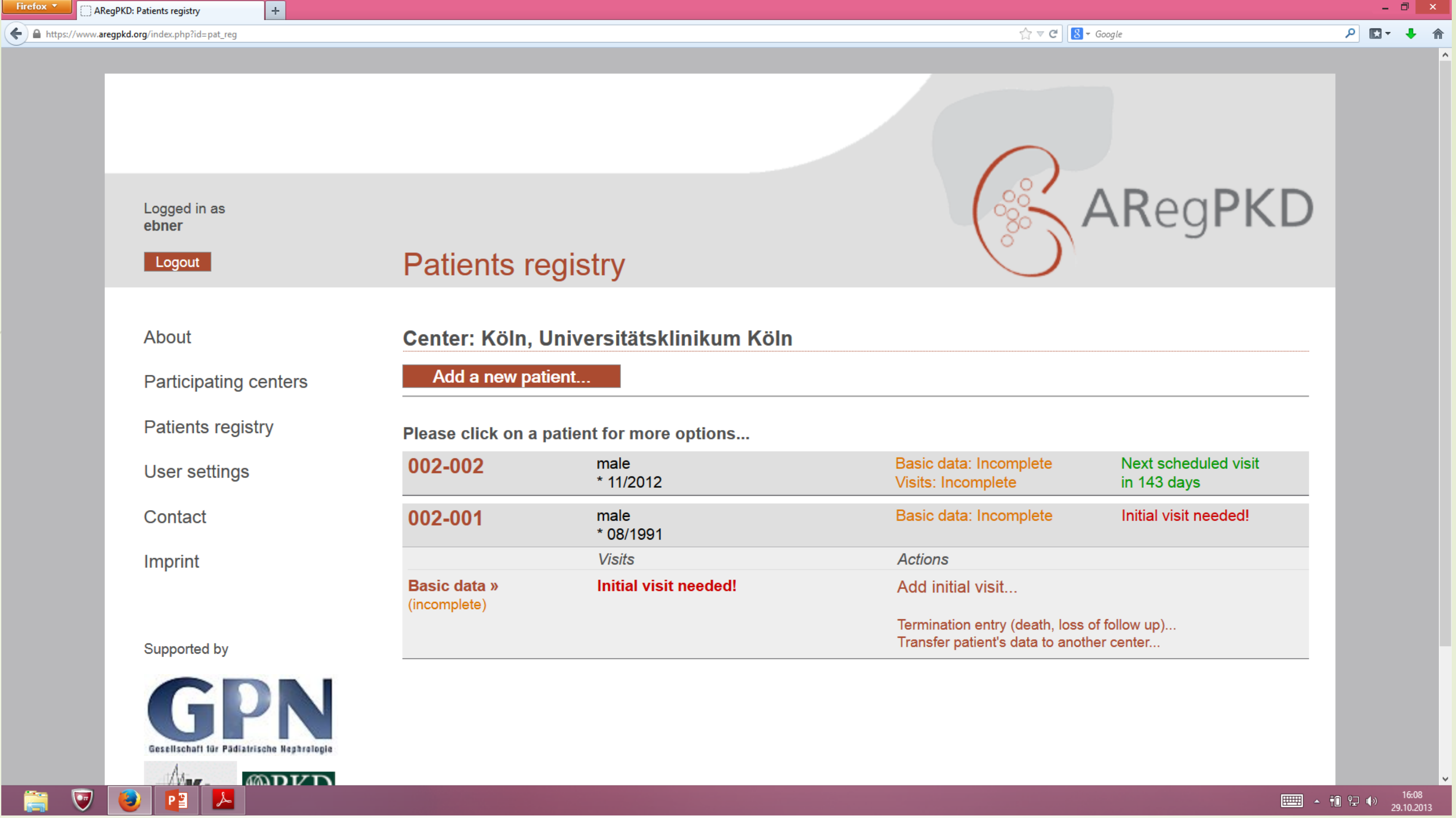
Important: If you want to add information to the *Basic Data* section later on you can always enter these topics in order to edit them.
For example: If your patient is receiving genetic testing you can enter the information in *Genetics* later on.



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Patients registry

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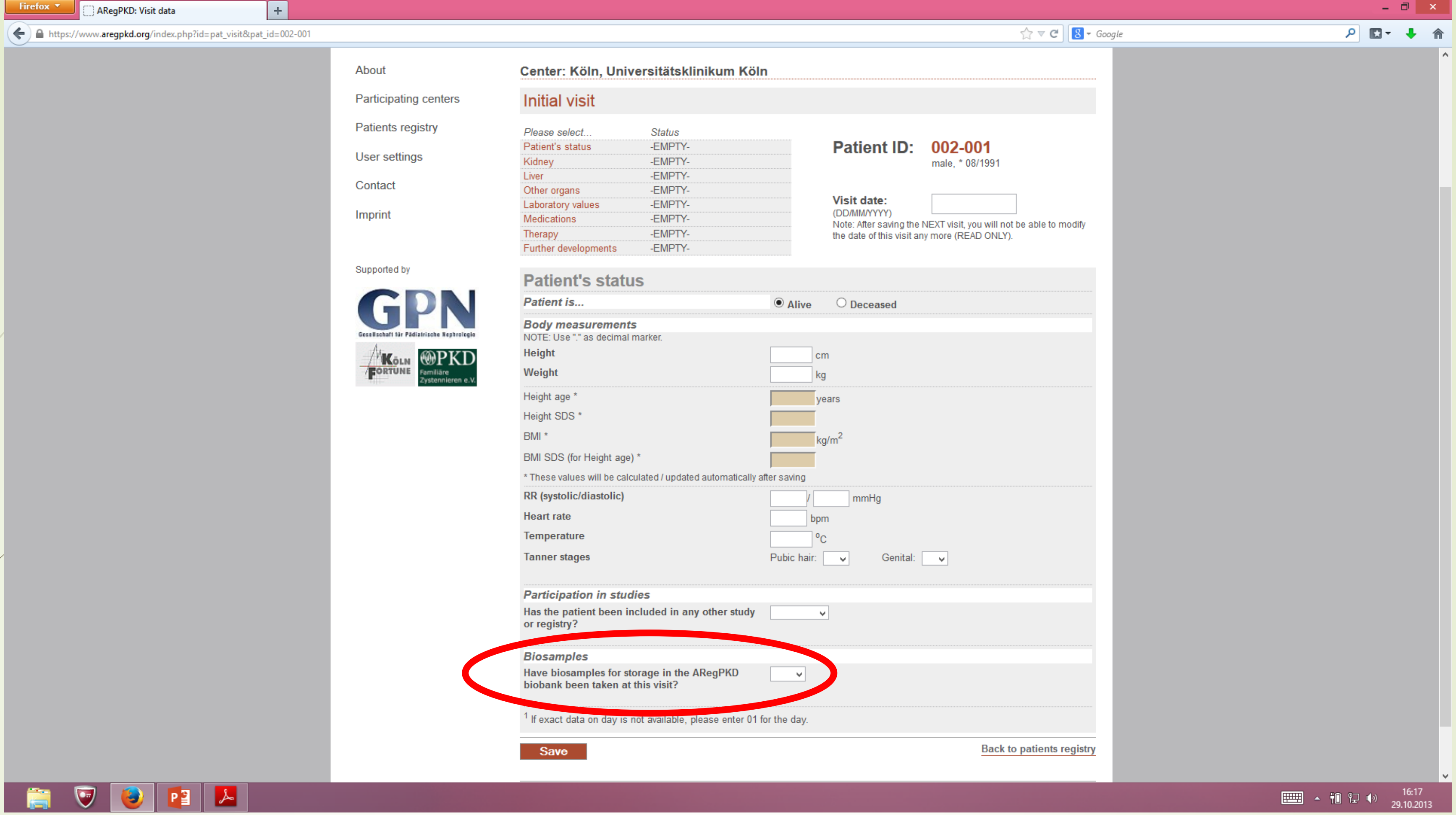
Please click on a patient for more options...

002-002	male * 11/2012	Basic data: Incomplete Visits: Incomplete	Next scheduled visit in 143 days
002-001	male * 08/1991	Basic data: Incomplete	Initial visit needed!
	Visits	*Actions*	
Basic data » (incomplete)	**Initial visit needed!**	Add initial visit... Termination entry (death, loss of follow up)... Transfer patient's data to another center...	

In your *Patients registry* you will be able to see whether a visit is needed.
Please enter data at least once every year.

Click on *Add initial visit* in order to fill in the information of the visit.
You can also fill in the *Termination entry* in case of death or loss of follow up or *Transfer patient`s data to another center*.



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Initial visit

Please select...	*Status*
Patient's status	-EMPTY-
Kidney	-EMPTY-
Liver	-EMPTY-
Other organs	-EMPTY-
Laboratory values	-EMPTY-
Medications	-EMPTY-
Therapy	-EMPTY-
Further developments	-EMPTY-

Patient ID: 002-001
male, * 08/1991

Visit date:
(DD/MM/YYYY)
Note: After saving the NEXT visit, you will not be able to modify the date of this visit any more (READ ONLY).

Patient's status

Patient is... ● Alive ○ Deceased

Body measurements

NOTE: Use "." as decimal marker.

Height cm
Weight kg

Height age * years
Height SDS *
BMI * kg/m^2
BMI SDS (for Height age) *

* These values will be calculated / updated automatically after saving

RR (systolic/diastolic) / mmHg
Heart rate bpm
Temperature °C
Tanner stages Pubic hair: Genital:

Participation in studies

Has the patient been included in any other study or registry?

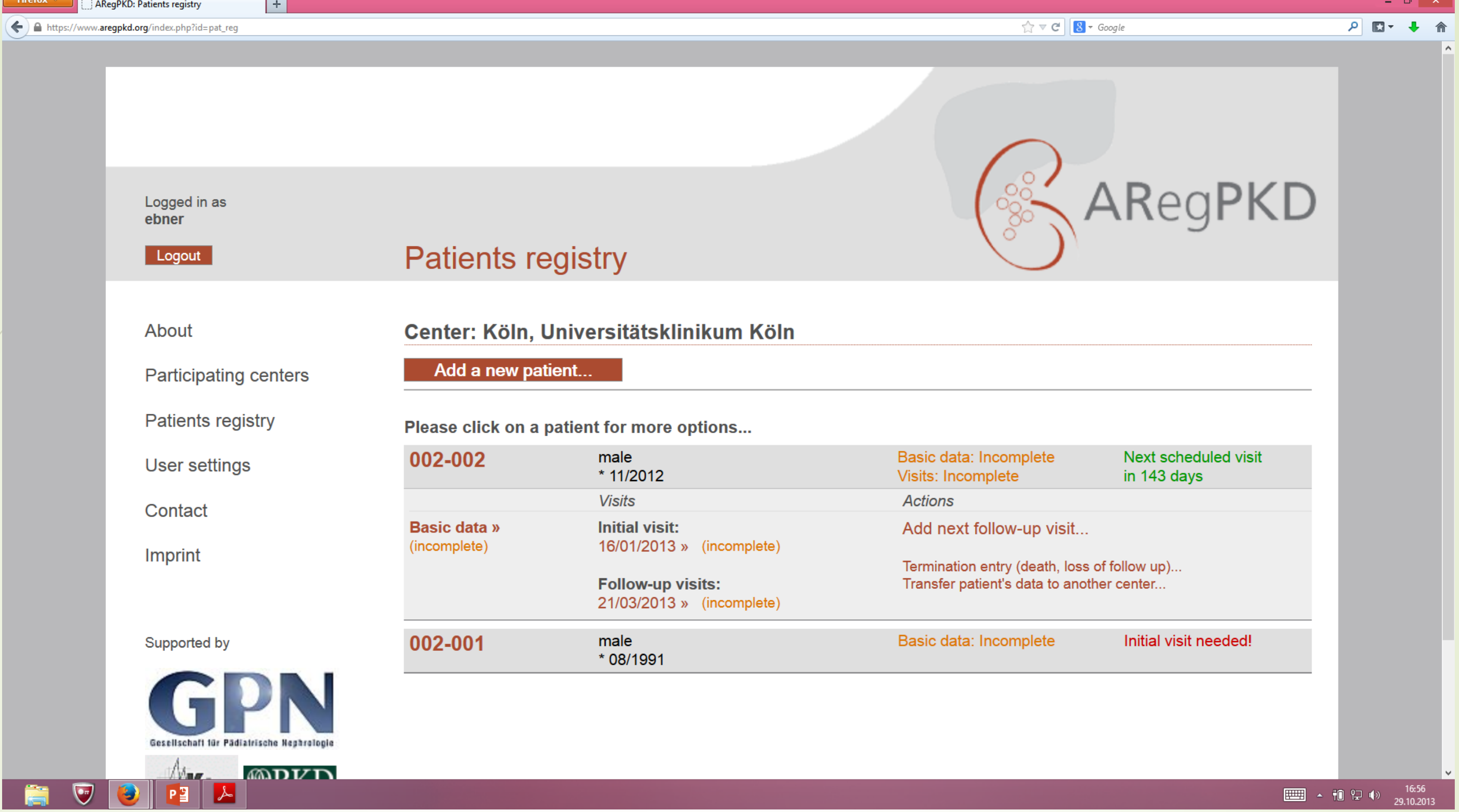
Biosamples

Have biosamples for storage in the ARegPKD biobank been taken at this visit?

[1] If exact data on day is not available, please enter 01 for the day.

You start the *Initial visit* with this view. The values in the orange fields will be calculated automatically after you pressed saving.

Please note: At the end of *Patient`s status* you are asked, if biosamples for storage in the ARegPKD biobank have been taken at this visit. We will send you corresponding labels for every year. If you have not yet received corresponding labels, please inform us!



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Logout

Patients registry

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Add a new patient...

Please click on a patient for more options...

002-002 male * 11/2012 Basic data: Incomplete Visits: Incomplete Next scheduled visit in 143 days

Visits *Actions*

Basic data » (incomplete)

Initial visit:
16/01/2013 » (incomplete)

Follow-up visits:
21/03/2013 » (incomplete)

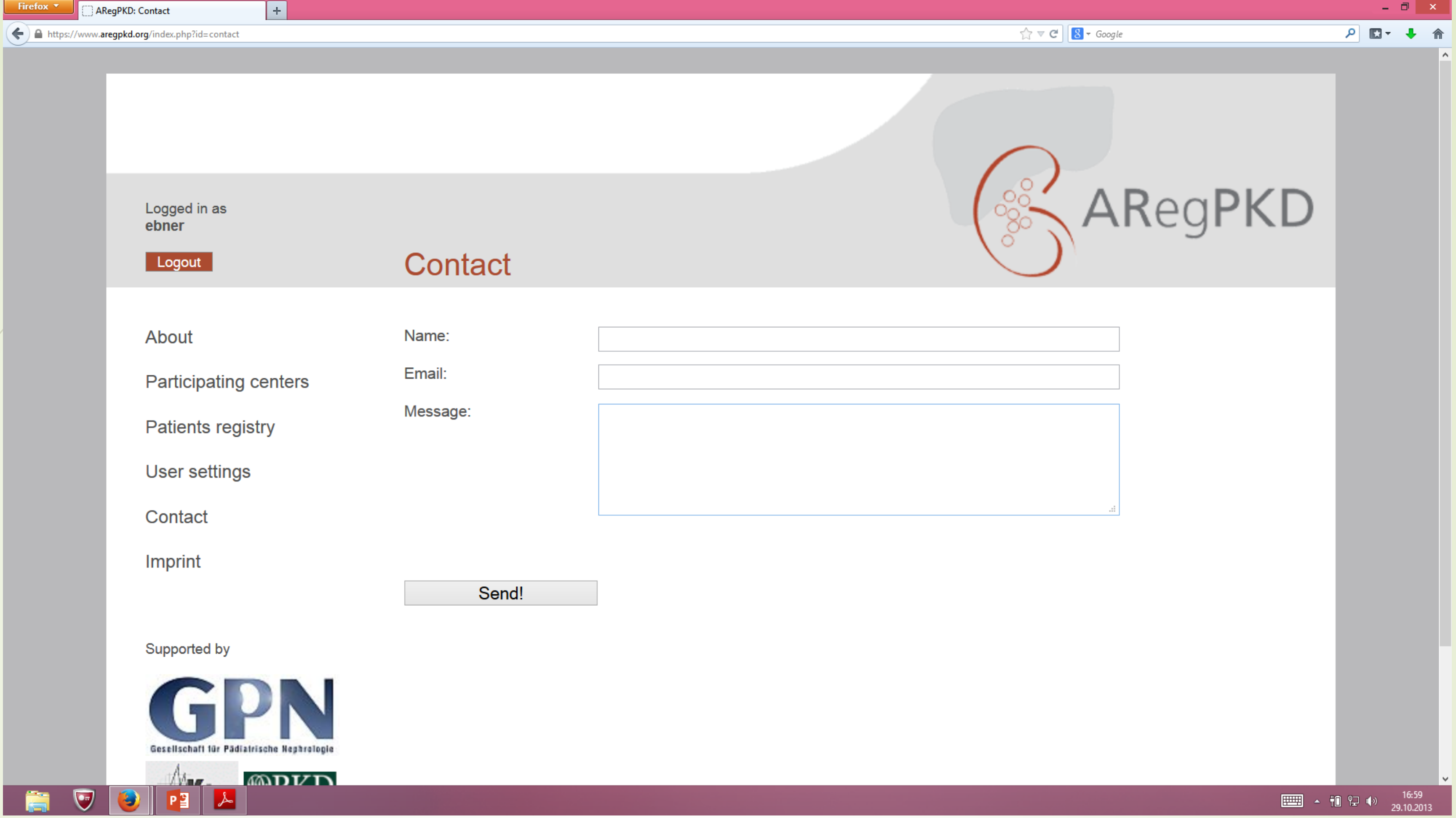
Add next follow-up visit...

Termination entry (death, loss of follow up)...
Transfer patient's data to another center...

002-001 male * 08/1991 Basic data: Incomplete Initial visit needed!

If your patient already has a documented *Initial visit*, you can progress with *Add follow-up visit* or *Add next follow-up visit*.

You also get an overview over all entered visits.



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Contact

Name:

Email:

Message:

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Whenever you have any questions regarding ARegPKD, please do not hesitate to contact us!

- Fill in the Contact form or
- Send an e-mail to info@aregpkd.org, kathrin.burgmaier@uk-koeln.de or max.liebau@uk-koeln.de