

# The PURA Syndrome Global Patient Registry



## *Getting Started* A User Guide

## **Before You Start**

### *Account Authorisation*

You will need to contact the Administrator of the PURA Syndrome Global Patient Registry to authorise your e-mail address for registration and access to the patient registry. To do this, contact [PURA@soton.ac.uk](mailto:PURA@soton.ac.uk)

Once you have received an acknowledgement from the Administrator, you will be able to use your authorised e-mail address to register your login credentials and access the patient registry through the web portal. **Please check your inbox and junk mail for an e-mail with the subject title 'PURA Patient Registry Account Activated'. You can use the embedded link to reach the patient registry.**

### *Clinical Documentation & Laboratory Reports*

It is strongly recommended that you have your child's laboratory report detailing their genetic diagnosis and any relevant clinical correspondence to hand when setting up your child's profile within the Patient Registry for the first time.

### *Duration*

Allow 30 minutes to:

- create your login credentials
- setup your child's profile
- read and sign the electronic consent form

There are currently 9 modules within the Patient Registry. Depending on the level of detail provided, these could take quite a long time to complete. We would like to collect the most accurate information that we can, so please take your time. You do not need to complete all of the modules at once. Even a partly completed module can be saved for another day.

## **Important Information about the Patient Registry**

The PURA Syndrome Global Patient Registry has been set up using an established secure healthcare database. There are limited options for development of the User Interface, which is aimed at healthcare professionals. Therefore, although as a parent or guardian you will be entering information about your child (who will appear as a 'participant' within the Patient Registry), there are some instances when you may be referred to as a 'clinician' or 'investigator' entering data on their behalf. Unfortunately, we cannot alter this terminology within the User Interface.

## **Support**

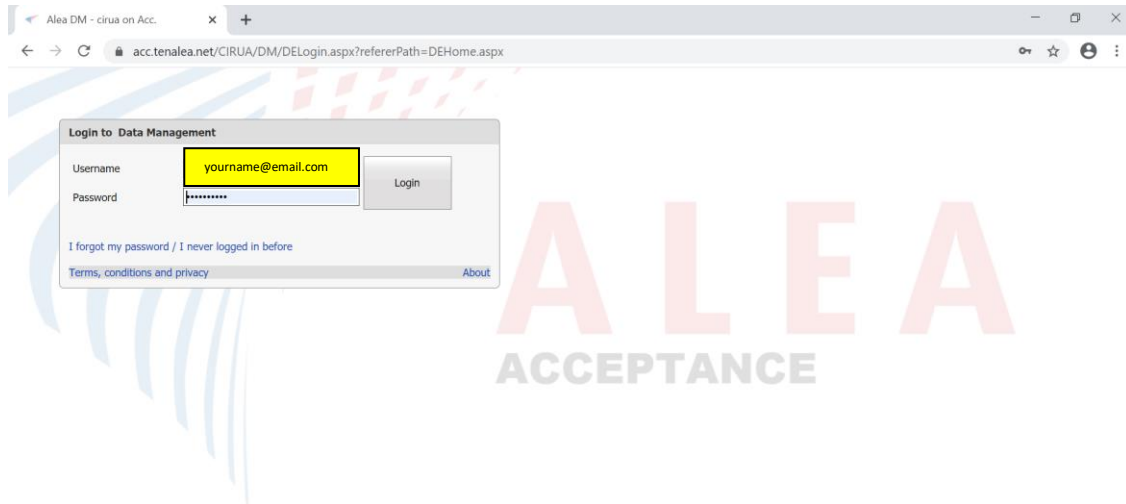
Hopefully, this step-by-step guide will help to make the navigation of the site as easy as possible. Once you have set up your child's profile within the Patient Registry, completion of the various linked questionnaires should be straightforward.

However, if you do experience any problems with the Patient Registry, please contact: [PURA@soton.ac.uk](mailto:PURA@soton.ac.uk)

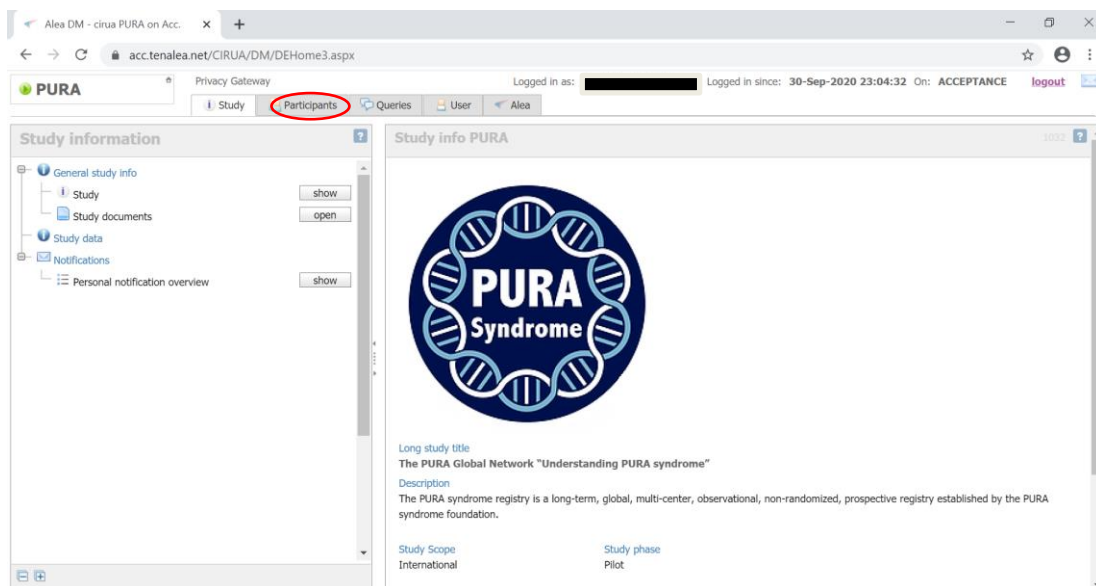


# The PURA Syndrome Global Patient Registry

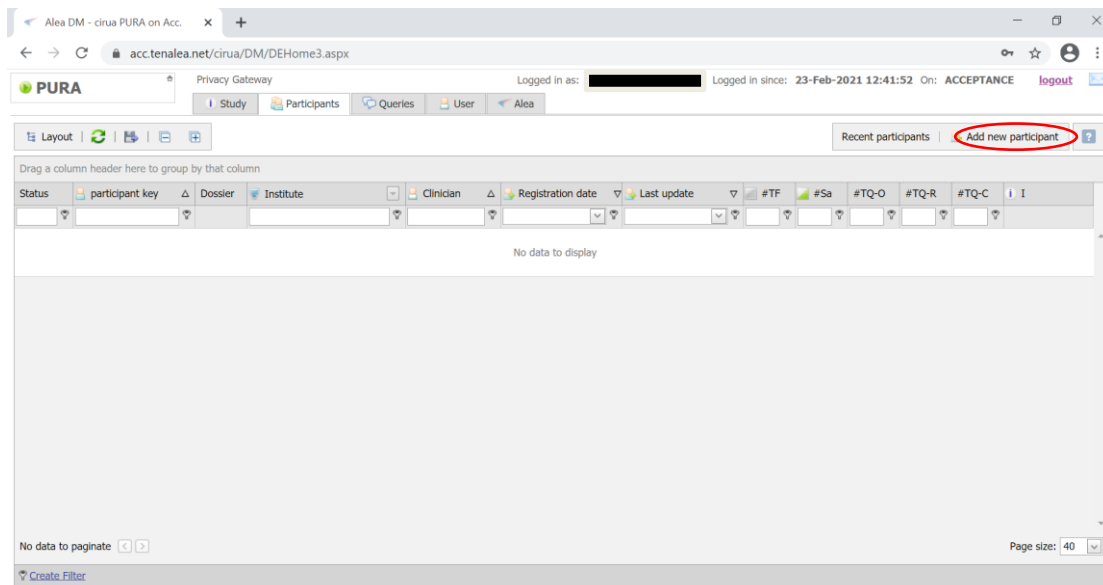
1. Navigate to the homepage: <https://prod.tenalea.net/ciru/DM/>
  - a. If this is the first time that you are using the site, click 'I never logged in before' and create your account using the e-mail address that has been authorised by the PURA patient registry administrator
  - b. If you have already created an account password, enter your credentials to login



2. Select the 'Participants' tab on the landing page

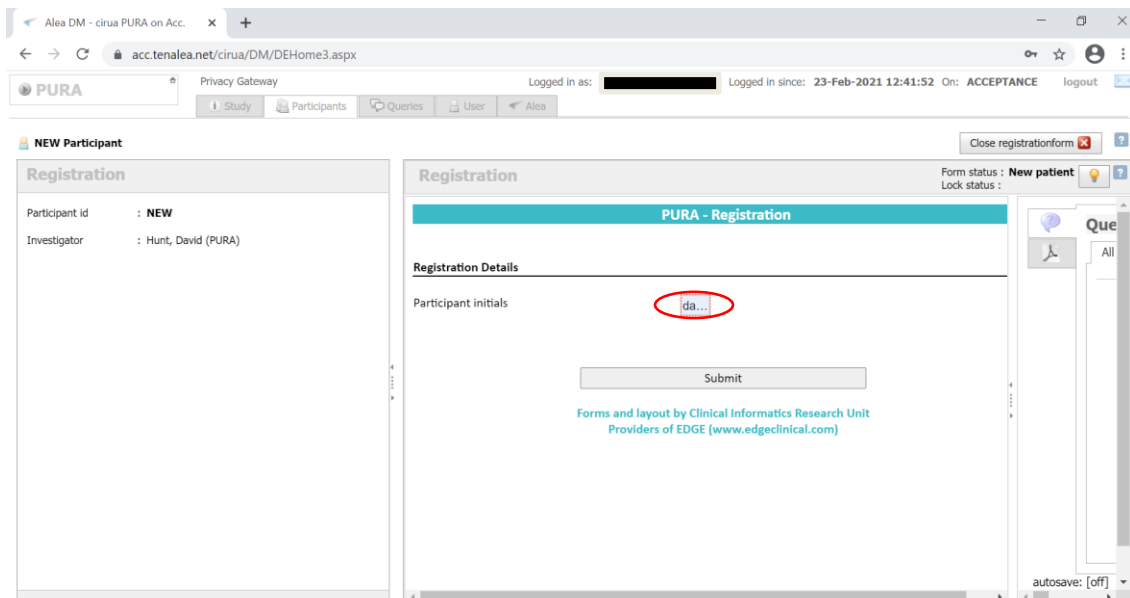


### 3. Click 'Add new participant'



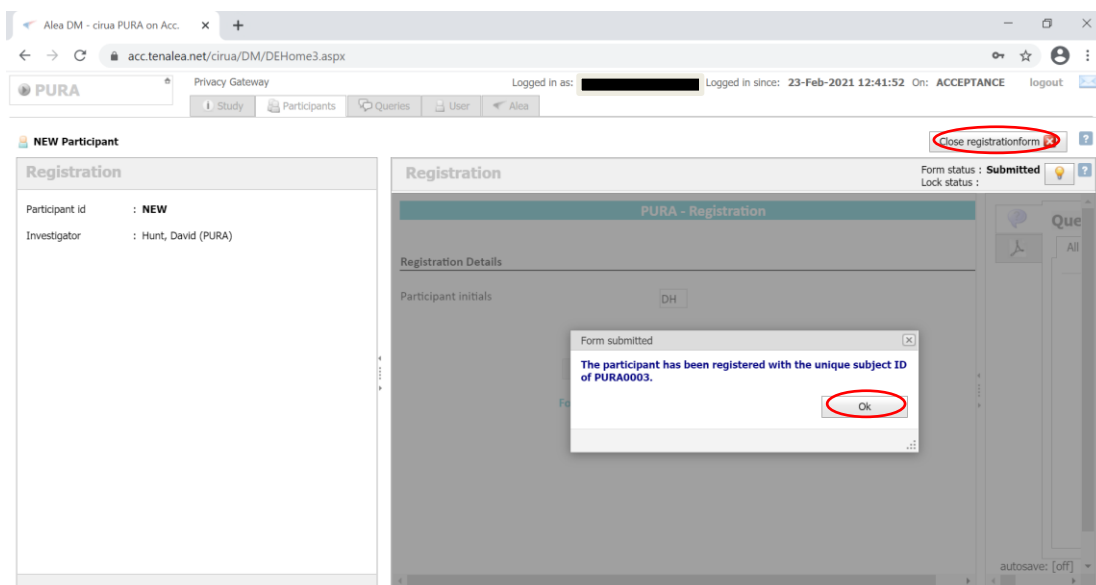
### 4. Add your child's initials in the 'participant initials' free text box, and then click 'Submit'

**N.B. If you need to register more than one child, please ensure that the initials that you assign to each child enable you to easily distinguish between them**

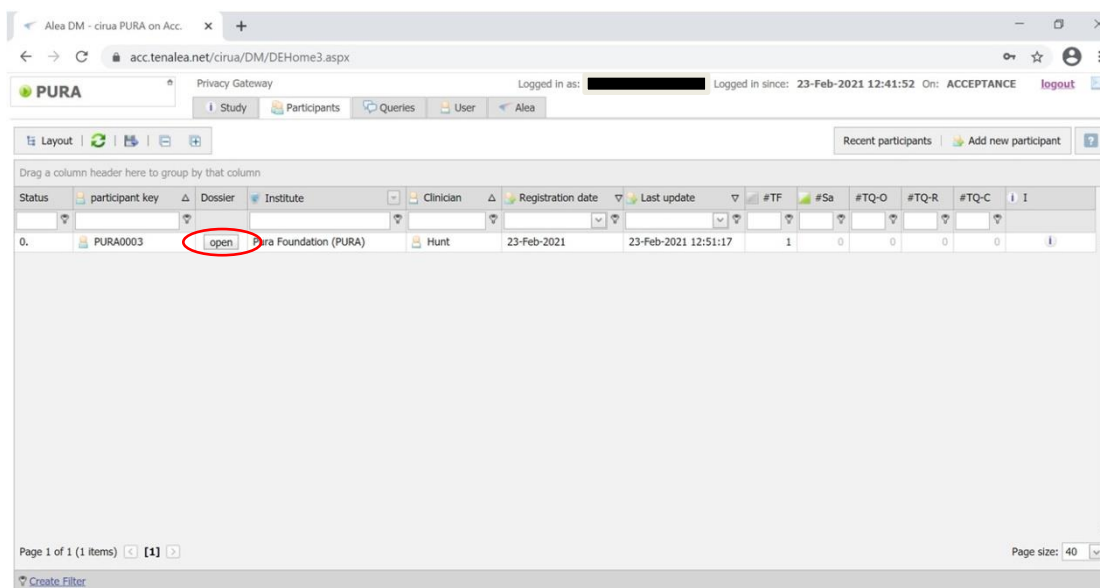


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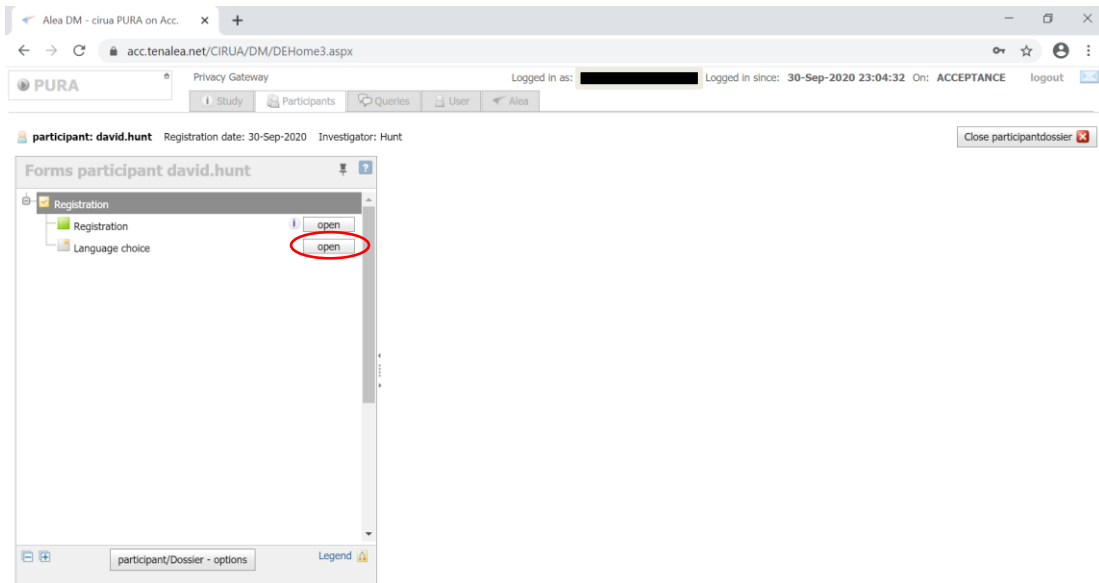
5. Click 'Ok' in the pop-up box to acknowledge the registration of the participant. Then click 'Close registrationform'



6. The participant details that you have entered should now be visible. Under the Dossier column, click 'Open'

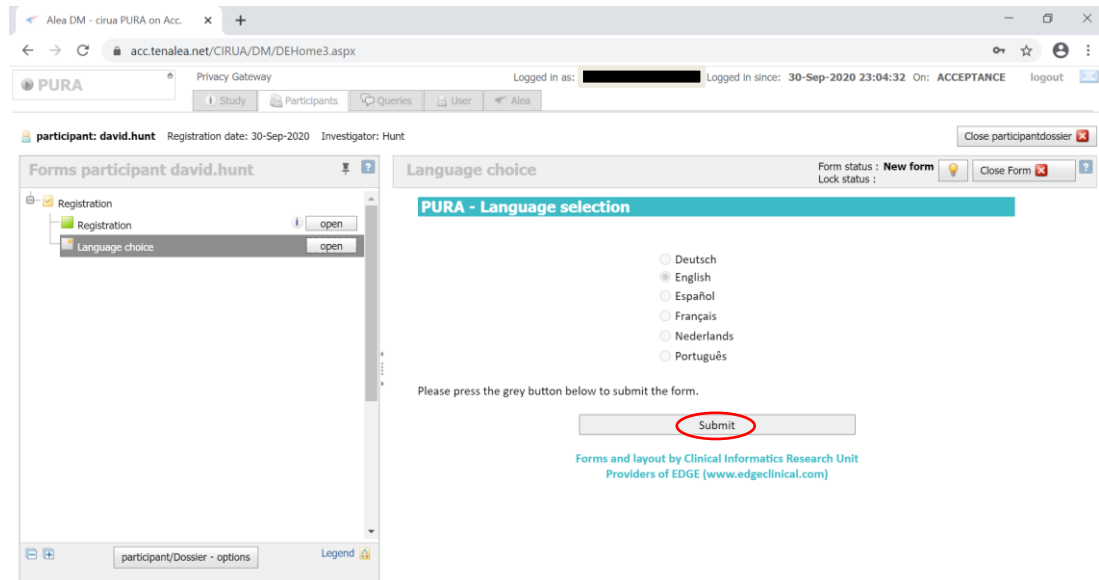


7. You will now see the participant forms screen. Click the 'Open' box, corresponding to Language choice

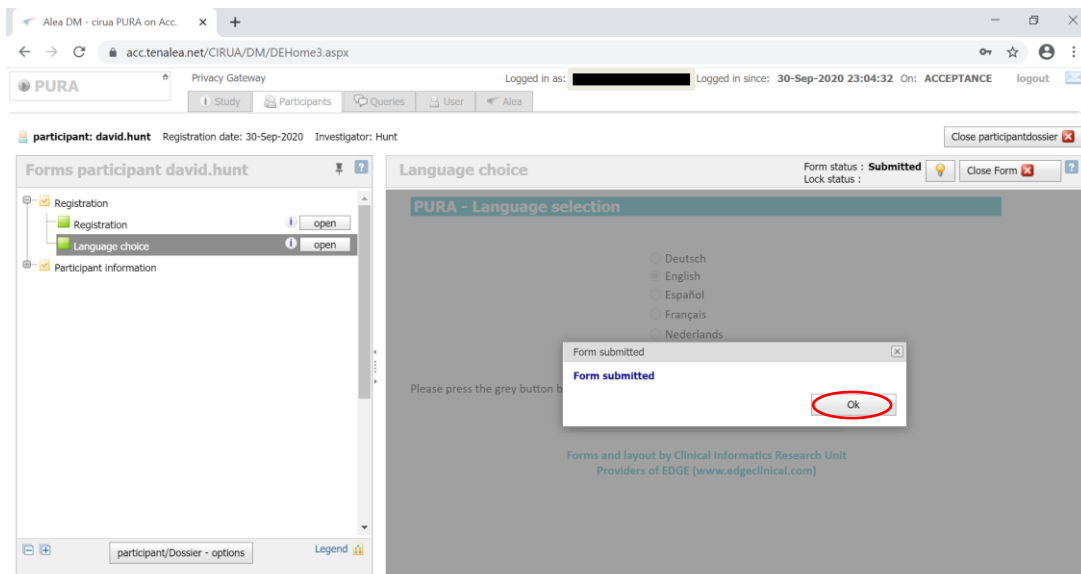


8. English is selected by default and there are currently no selectable options for other languages. Click 'Submit'.

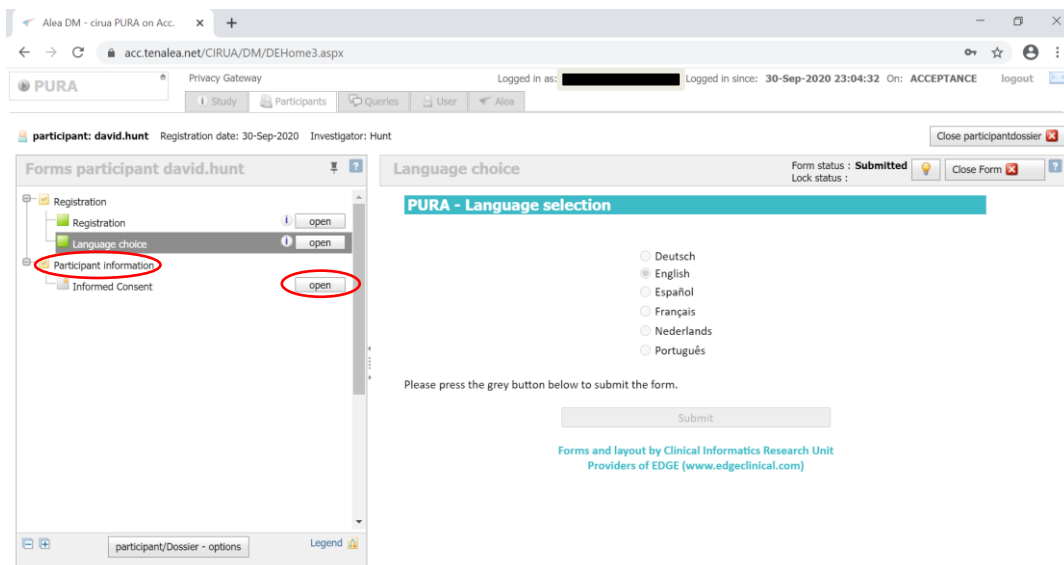
**N.B. We hope to make other languages available in future**



- Click 'OK' in the pop-up box to acknowledge the form submission. Note that the Language choice option is now marked green



- First click on 'Participant information', then click the 'Open' box, corresponding to Informed consent.



## 11. Read the consent form and enter **your initials** in each free text box if you agree with the accompanying statement

Forms participant david.hunt

Informed Consent

PURA - Consent Form - Understanding PURA Syndrome [V2.0, Sept 2018]

Southampton University Hospital Southampton NHS

The PURA Global Network "Understanding PURA syndrome"

		Initial each statement if you agree
1	I confirm that I have parental responsibility/legal guardianship and am the main custodian.	D-H
2	I confirm that I have read and understand the information sheet (version 1.0) for the above study and have had the opportunity to ask questions.	D-H
3	I understand that participation is voluntary and that I am free to withdraw my child at any time, without giving any reason and without medical care or legal rights being affected.	dh
4	In the event that I withdraw my child from the study, I understand that data already submitted will remain in the study but that no further data will be requested.	
5	I understand access to relevant information contained within my child's medical records is required in order to conduct this research project.	
6	I understand that data will be held subject to the regulations on data protection (General	

## 12. Only when initials are provided in all 11 fields does the submit button appear at the bottom of the form. Click 'Submit'.

Forms participant david.hunt

Informed Consent

policy). Personal and medical information will be encrypted and stored on a secure server in Den Bosch, the Netherlands. Access to my child's data is strictly limited to authorised users.

7	I understand that all data will be handled confidentially and that any information that could identify my child will be removed if the data is published, unless explicit written consent is first obtained.	D-H
8	I agree to the sharing of my child's anonymised data with national and international research collaborators.	D-H
9	I am happy to be contacted by the research team during the study period.	D-H
10	I am happy to be contacted about future studies relevant to my child's condition. I understand there will be no obligation to take part.	D-H
11	I agree to my child taking part in the above study.	D-H

The submit button will become enabled when at least items 1-7 and 11 have been initialed, all initials are the same and only contains alpha-character in the format ABC or A-C.

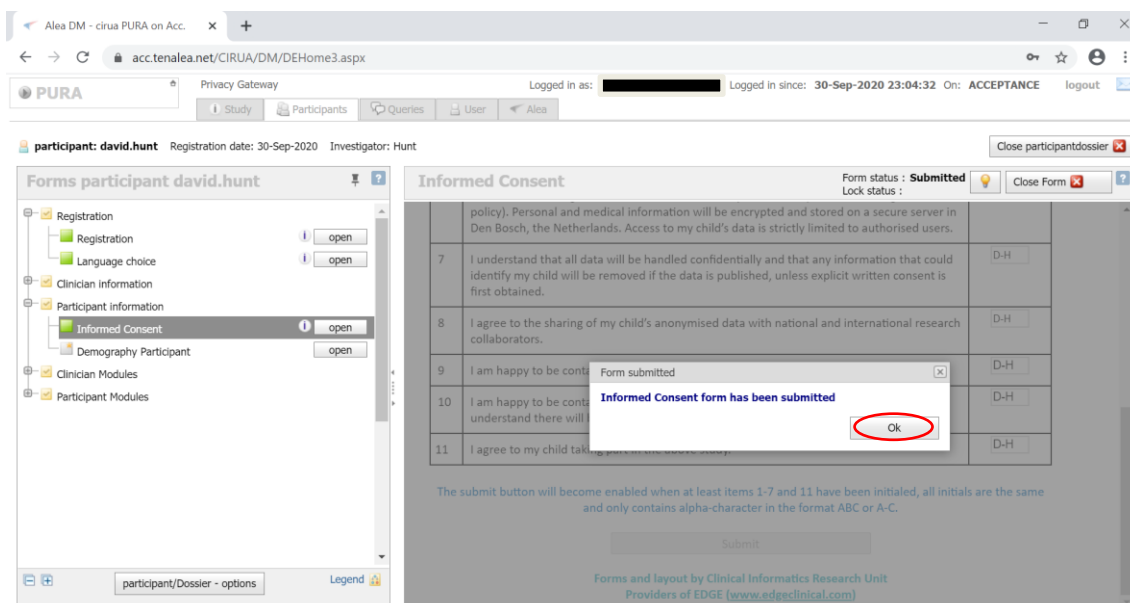
Submit

Forms and layout by Clinical Informatics Research Unit  
Providers of EDGE ([www.edgeclinical.com](http://www.edgeclinical.com))



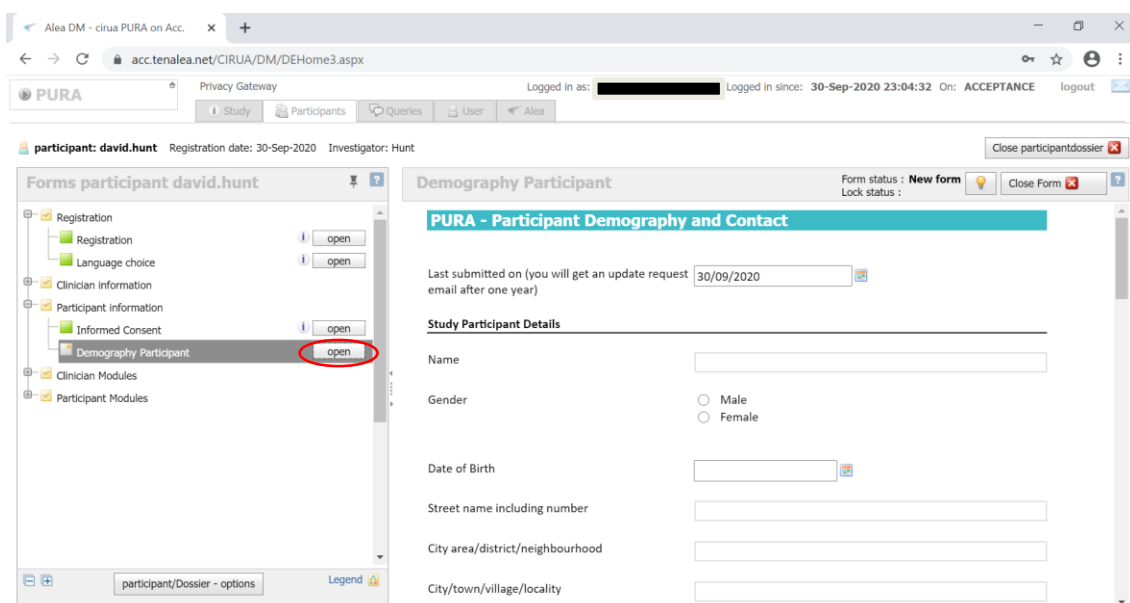


13. Click 'OK' in the pop-up box to acknowledge the submission of the consent form. Note that Informed consent is now marked green in the left-hand pane.



14. Click the 'Open' box, corresponding to Demography Participant. Please note that completion of this form is optional.

**N.B. Although this is optional, we would strongly encourage you to at least provide the following information: gender, DOB, county/state/country and genetic diagnosis.**



## 15. Next, expand the Participant Modules option

The screenshot shows a web browser window with the URL `acc.tenalea.net/CIRUA/DM/DEHome3.aspx`. The page title is "PURA" and it shows a "Privacy Gateway" and "Logged in as: [redacted]". The main content area is titled "Forms participant david.hunt" and "Demography Participant". The left-hand navigation menu is expanded to show "Participant Modules" with a red circle around it. The "Participant Modules" menu includes the following options: M1P2 - Genetics and Family History, M2P2 - Pregnancy, Birth and Newborn, M3P2 - Developmental Milestones Part 1, M3P2 - Developmental Milestones Part 2, M4P2 - Neurological History, M5P2 - Seizures and Epilepsy, M6P2 - Growth, and M7P2 - Endocrine. The "Demography Participant" form is visible on the right, showing fields for Name, Gender, Date of Birth, Street name including number, City area/district/neighbourhood, and City/town/village/locality.

## 16. Click the 'Open' box, corresponding to M1P2 - Genetics and Family History

The screenshot shows the same web browser window as above, but now the "M1P2 - Genetics and Family History" form is open in the left-hand navigation menu, with a red circle around it. The main content area is titled "M1P2 - Genetics and Family History" and "PURA - Participants - Module 1: Genetic and Family History". The form contains a "Genetic Diagnosis" section with three questions:

- How old was your child when he/she was diagnosed?
  - One year or older:  years
  - Under one year:  months
  - Not sure
- How old was your child when they started having genetic tests?
  - One year or older:  years
  - Under one year:  months
  - Not sure
- What condition was your child diagnosed with?
  - PURA syndrome
  - 5q.31.3 deletion syndrome including PURA
  - 5q.31.3 duplication syndrome including PURA
  - Not sure



17. The first module, Genetics and Family History, is now visible. This can be completed using a combination of radio buttons, drop down options and free-text fields.

The screenshot shows a web browser window with the URL `acc.tenalea.net/CIRUA/DM/DEHome3.aspx`. The page title is "PURA". The user is logged in as "Alea" and the participant is "david.hunt". The form is titled "M1P2 - Genetics and Family History" and is in "New form" status. The form content includes:

- Genetic Diagnosis**
- Question 1: "How old was your child when he/she was diagnosed?" with radio buttons for "One year or older" (with a dropdown for years), "Under one year" (with a dropdown for months, set to 7), and "Nor sure".
- Question 2: "How old was your child when they started having genetic tests?" with radio buttons for "One year or older" (with a dropdown for years), "Under one year" (with a dropdown for months, set to 4), and "Not sure".
- Question 3: "What condition was your child diagnosed with?" with radio buttons for "PURA syndrome", "5q.31.3 deletion syndrome including PURA", "5q.31.3 duplication syndrome including PURA", and "Not sure".

18. Scroll to the bottom of the page and click 'submit' to save the form (this can be done, even if the form is incomplete)

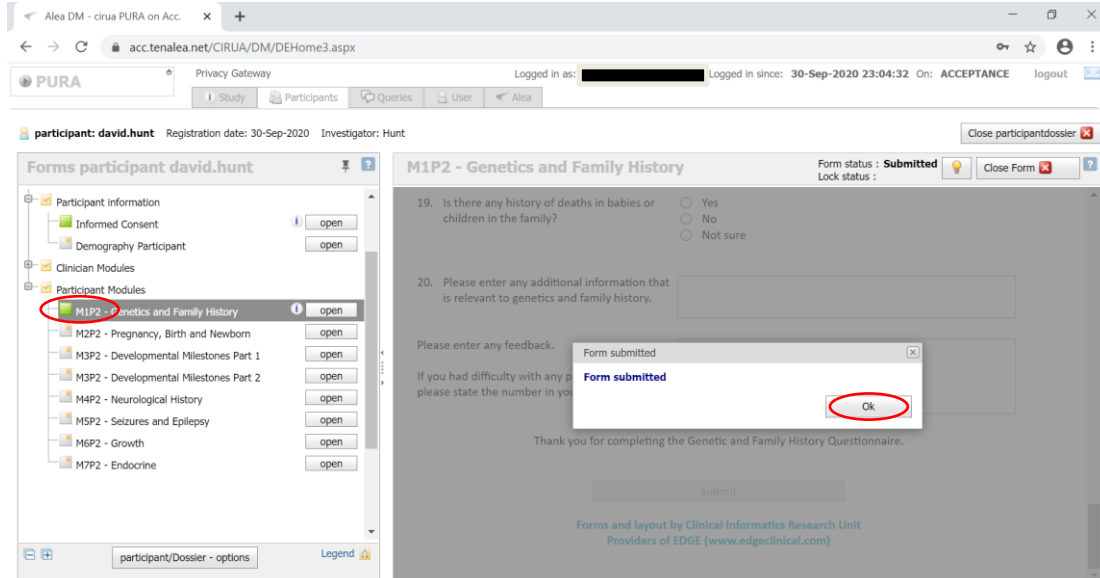
The screenshot shows the bottom of the "M1P2 - Genetics and Family History" form. The form content includes:

- Question 19: "Is there any history of deaths in babies or children in the family?" with radio buttons for "Yes", "No", and "Not sure".
- Question 20: "Please enter any additional information that is relevant to genetics and family history." with a text input field.
- A feedback section: "Please enter any feedback." with a text input field and the instruction "If you had difficulty with any particular question, please state the number in your feedback."
- A "Thank you for completing the Genetic and Family History Questionnaire." message.
- A "Submit" button, which is circled in red.
- Footer text: "Forms and layout by Clinical Informatics Research Unit Providers of EDGE ([www.edgeclinical.com](http://www.edgeclinical.com))"



19. Click 'OK' in the pop up box to acknowledge submission of the form.  
Note that the Genetics and Family History tab is now marked green.

**N.B. The green tab does not mean that the form is locked. It can still be edited.**

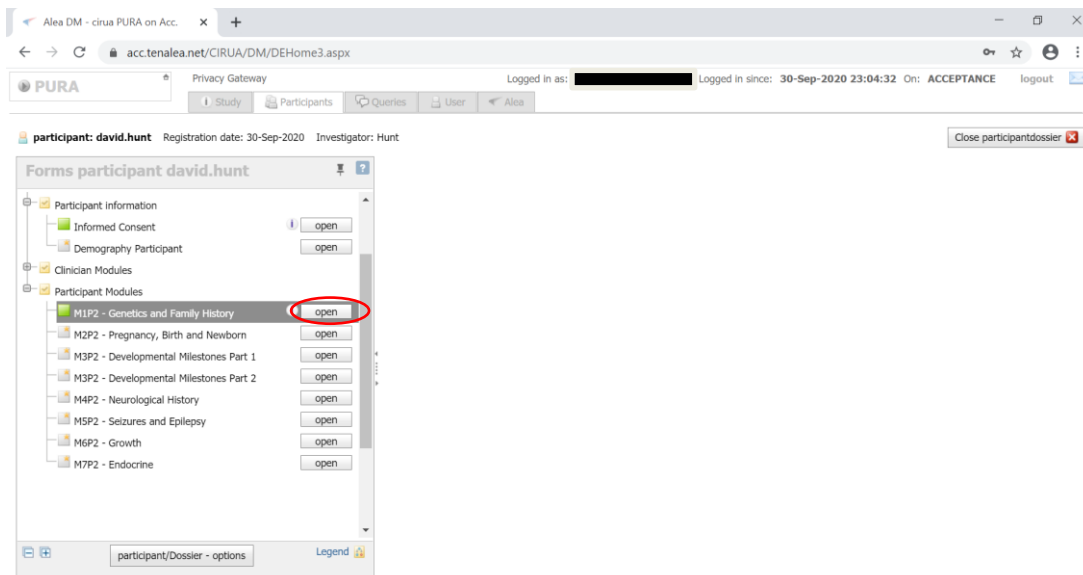


*You can now go through the same process to complete the remaining modules*

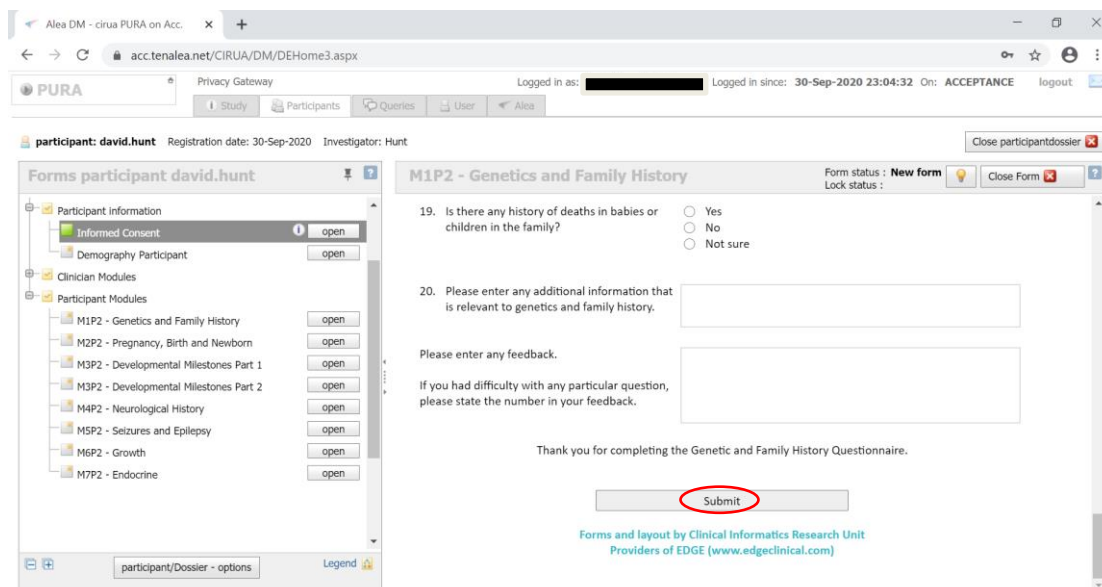


## Editing Participant Modules

1. Select the 'Open' box for the form that you wish to edit (any forms for which data is already stored are marked green).



2. Edit the form as appropriate and then click 'Submit' at the bottom of the form.



## Exiting a form without saving any changes

1. If you wish to close the form without saving any data click 'Close Form' instead.

The screenshot shows a web browser window with the URL `acc.tenalea.net/CIRUA/DM/DEHome3.aspx`. The user is logged in as 'Alea DM - cirua PURA on Acc.'. The page displays a participant dossier for 'david.hunt' with a registration date of '30-Sep-2020'. The left sidebar shows a tree view of modules, with 'M1P2 - Genetics and Family History' selected. The main content area shows the form 'M1P2 - Genetics and Family History' with a status of 'Submitted'. A red circle highlights the 'Close Form' button in the top right corner of the form area.

2. In the pop up that warns that closing the form may lose unsaved data, only click 'OK' if you are certain that you wish to exit without saving any data. Otherwise, click 'cancel' to return to the form.

The screenshot shows the same web browser window as above, but with a confirmation dialog box overlaid. The dialog box text reads: "acc.tenalea.net says This form may contain unsaved data. Continue closing anyway?". The 'OK' button is highlighted with a red circle.



## Clinician Modules

- Note that the Clinician Modules cannot be created or edited by parents. However, once these have been completed by your child's clinician, they will be visible to you in read only form.

The screenshot shows a web browser window with the URL `acc.tenalea.net/CIRUA/DM/DEHome3.aspx`. The page title is "Forms participant david.hunt". The navigation bar includes "Study", "Participants", "Queries", "User", and "Alea". The user is logged in as "Alea" and the session is from "30-Sep-2020 23:04:32". The main content area displays a tree view of forms for participant "david.hunt". The "Clinician Modules" section is highlighted with a red circle and contains the following items:

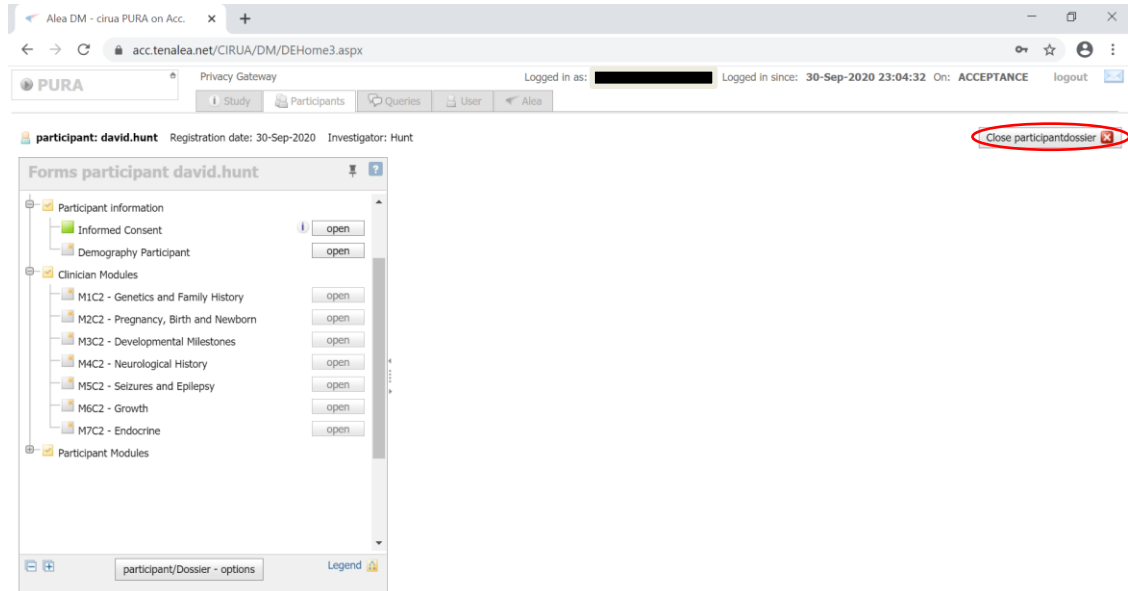
- M1C2 - Genetics and Family History (open)
- M2C2 - Pregnancy, Birth and Newborn (open)
- M3C2 - Developmental Milestones (open)
- M4C2 - Neurological History (open)
- M5C2 - Seizures and Epilepsy (open)
- M6C2 - Growth (open)
- M7C2 - Endocrine (open)

Other sections include "Participant Information" (Informed Consent, Demography Participant) and "Participant Modules". A "Close participantdossier" button is visible in the top right corner.

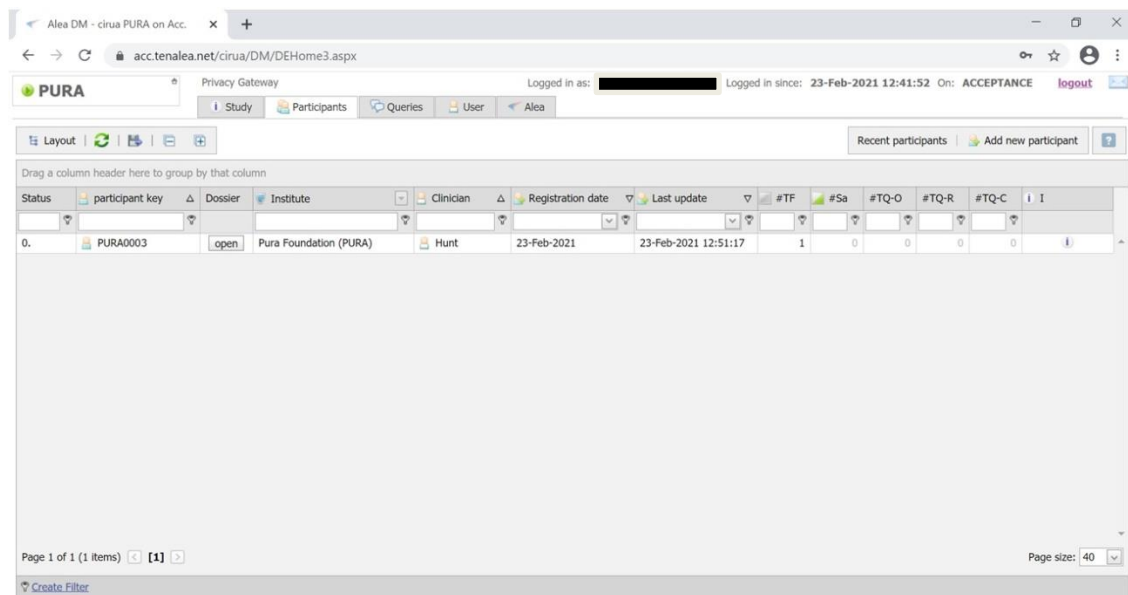


## Returning to Participant list

1. Click the 'Close participantdossier' button to return to the participant list.



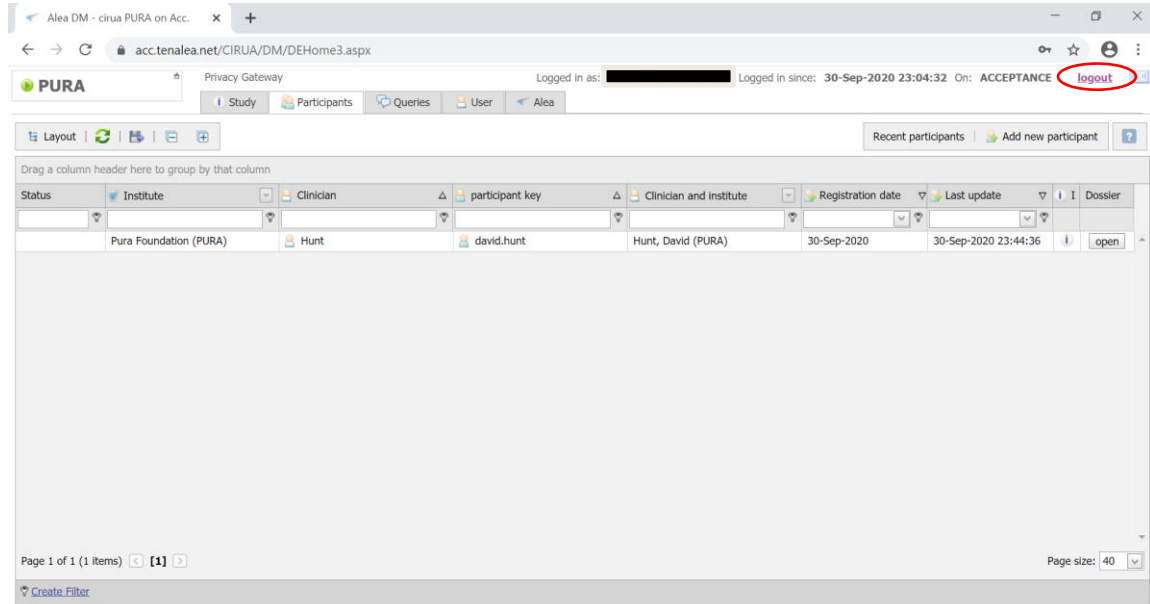
2. The following screen will be displayed:





## Logging out

- Click 'logout' to exit the patient registry



The screenshot shows a web browser window with the URL `acc.tenalea.net/CIRUA/DM/DEHome3.aspx`. The user is logged in as 'ALEA' and the session is labeled 'ACCEPTANCE'. A red circle highlights the 'logout' button in the top right corner of the application. The main content area displays a table of participants with the following columns: Status, Institute, Clinician, participant key, Clinician and institute, Registration date, Last update, and Dossier. A single participant is listed: Pura Foundation (PURA), Hunt, david.hunt, Hunt, David (PURA), 30-Sep-2020, 30-Sep-2020 23:44:36, and an 'open' link. The page footer indicates 'Page 1 of 1 (1 items)' and 'Page size: 40'.

Status	Institute	Clinician	participant key	Clinician and institute	Registration date	Last update	Dossier
	Pura Foundation (PURA)	Hunt	david.hunt	Hunt, David (PURA)	30-Sep-2020	30-Sep-2020 23:44:36	open

