

Manual for Submitting Clinical Data to the ECARUCA Database

After a case has been submitted by the cytogeneticist, the case record is available for receiving clinical data. Submitting of clinical data is usually done by the clinician involved in a particular case, but may also be performed by the cytogeneticist if he or she has the clinical details.

You need an ECARUCA clinician account to be able to submit clinical information. An account request form is available online at the log in page.

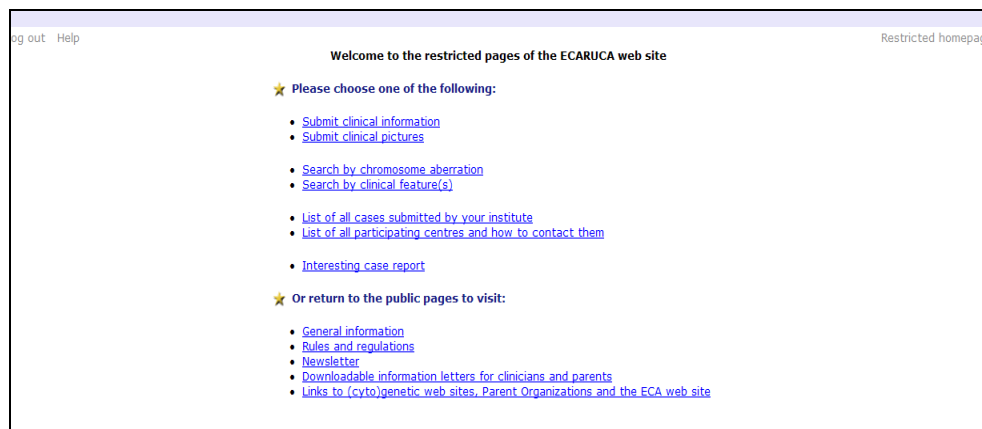
Follow the steps below to submit clinical data to the ECARUCA database:

1. Log in with your clinician account



The screenshot shows the ECARUCA website interface. On the left, a navigation menu includes 'Member' (circled in red) and 'Log in'. The main content area features a 'Log in' section with a form for 'Account name' (containing 'user') and 'Password' (masked with dots). A blue 'Log in' button is positioned to the right of the password field. Below the form, there are links for 'Password forgotten? Please [contact us](#) .' and 'Not an ECARUCA-member yet? Please go to the [account request form](#) .' The top of the page displays the ECARUCA logo and a header with three small portraits. The bottom of the page contains a small note about chromosome coordinates: 'Current coordinates are hg17, hg18 or hg19 depending on the microarray platform used. Please take this into account when searching the database via base position.'

You will access the restricted pages of ECARUCA



The screenshot shows the 'Restricted homepage' of the ECARUCA website. The page title is 'Welcome to the restricted pages of the ECARUCA web site'. It contains two main sections of links:

- Please choose one of the following:**
 - [Submit clinical information](#)
 - [Submit clinical pictures](#)
 - [Search by chromosome aberration](#)
 - [Search by clinical feature\(s\)](#)
 - [List of all cases submitted by your institute](#)
 - [List of all participating centres and how to contact them](#)
 - [Interesting case report](#)
- Or return to the public pages to visit:**
 - [General information](#)
 - [Rules and regulations](#)
 - [Newsletter](#)
 - [Downloadable information letters for clinicians and parents](#)
 - [Links to \(cyto\)genetic web sites, Parent Organizations and the ECA web site](#)

2. Click on the link 'Submit clinical information'

log out Help Restricted homepage

Welcome to the restricted pages of the ECARUCA web site

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ECARUCA shows all cases ready for submission on your account

Registration

Here are your cases available for the submission of clinical data. Please select the case you wish to register:

| case id | dob | country | gender |
|---------|------------|-----------------|--------|
| 4569 | 2001-01-26 | The Netherlands | Female |
| 4568 | 2007-10-30 | The Netherlands | Female |
| 4440 | 2004-02-12 | The Netherlands | Male |

To run the above Java applet, you will require [the Sun Java plugin](#) (at least version 1.4.0_01), and IE 6 or Firefox/Netscape 7 (or higher). See [FAQ](#) page for further details.

3. Select the case you wish to register

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4. Enter the clinical data

Case Detail

Case ID: 1956 DOB: 1958-05-09 Country: The Netherlands Gender: Female

Clinical Data Clinical Features Other Clinical Investigations Pedigree Data Publications Submission

Age at Last Clinical Examination: Year Month

Mental Retardation Level:

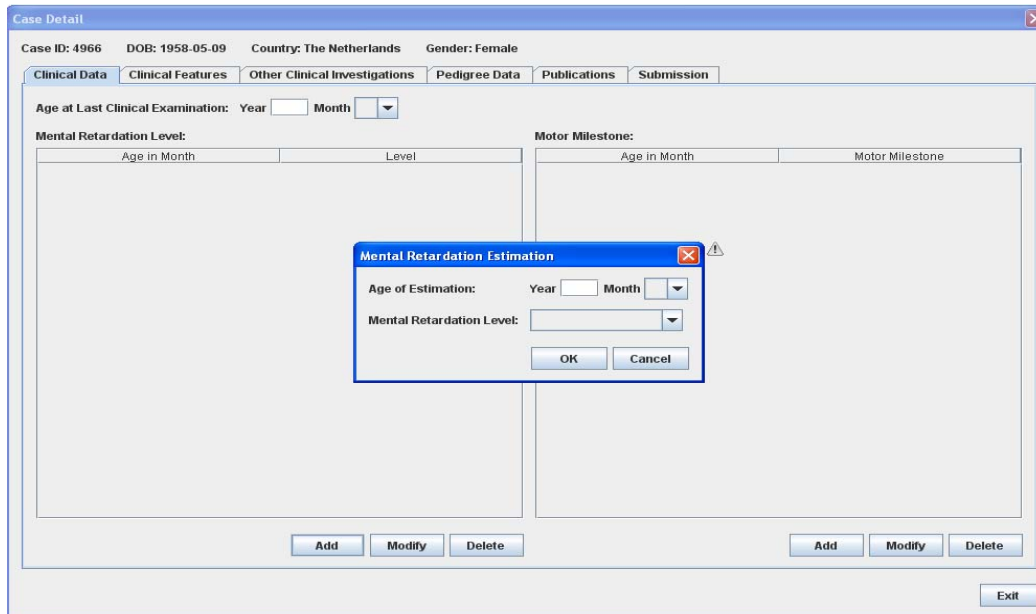
| Age in Month | Level |
|---------------|-------|
| Clinical data | |

Motor Milestone:

| Age in Month | Motor Milestone |
|---------------|-----------------|
| Clinical data | |

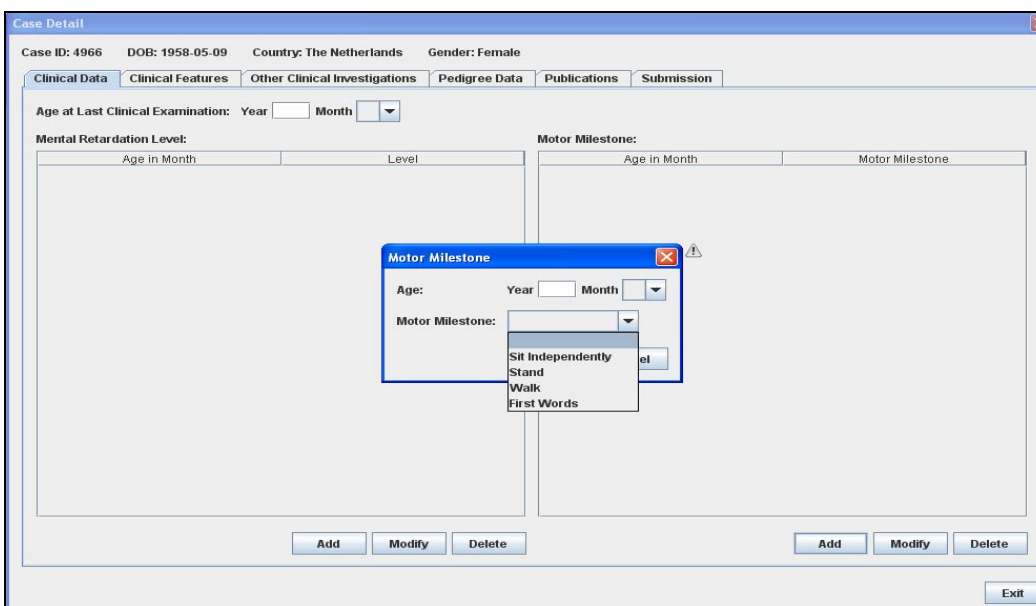
- Enter the year and choose the month of the Age at Last Clinical Examination. If the age of the patient is less than 1 month, enter zero in both fields.

- Enter the level of mental retardation: Press the add button to enter the level of mental retardation. Enter the required information and press the 'OK' button.



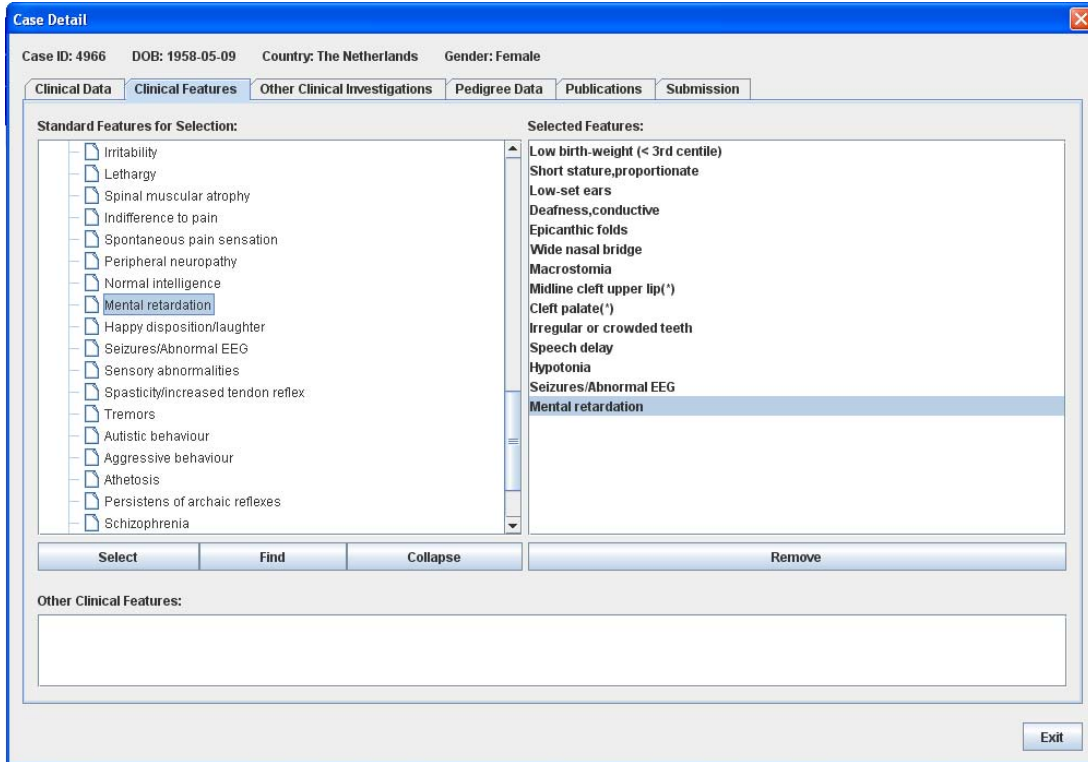
The information is now displayed on the Clinical Data tab. Press the button 'modify' if you wish to change the entry or press the button 'delete' if you want to remove the entry.

- Enter the milestones. Press the add button to enter the different milestones. Enter the required information and press 'OK'.



The information is now displayed on the Clinical Data tab. Press the button 'modify' if you wish to change the entry or press the button 'delete' if you want to remove the entry.

5. Enter Clinical Features.



In the section Standard Features, an expandable tree of clinical features is shown. The tree of features is a hierarchical structure in which the features are divided into (sub)categories. A description of a feature is obtained by right-clicking on the feature of choice.

There are two ways of finding a specific clinical feature:

- Expand the tree by double-clicking on the category (and sub-categories if applicable) until the desired feature is listed.
- Search for a feature by pressing the "find" button, the Feature Search window is displayed.

When you have found a clinical feature, add it to the case by clicking this feature and the select button thereafter. The selected features will be visible in the Selected Features tab. Features which are not listed in the feature tree can be added to the case by entering the description in the field "Other Clinical Features".

If you wish to remove features, select the specific feature and click the 'remove' button.

6. Enter Other Clinical Investigations

Click on the tab “Other Clinical Investigations” if you want to record additional information. Press the button “Add”, the window “Other Clinical Investigation” displays.

The screenshot shows a software interface for entering clinical data. The main window is titled "Case Detail" and displays patient information: Case ID: 4966, DOB: 1958-05-09, Country: The Netherlands, Gender: Female. It has several tabs: Clinical Data, Clinical Features, Other Clinical Investigations (selected), Pedigree Data, Publications, and Submission. A table with columns "Investigation", "Date", and "Result" is visible. A modal dialog box titled "Other Clinical Investigation" is open, containing the following fields:

- Standard Investigation Test: (dropdown menu)
- Other Investigation Test: (text input field)
- Test Result: (text input field)
- Test Date: (Year, Month, Day dropdowns)

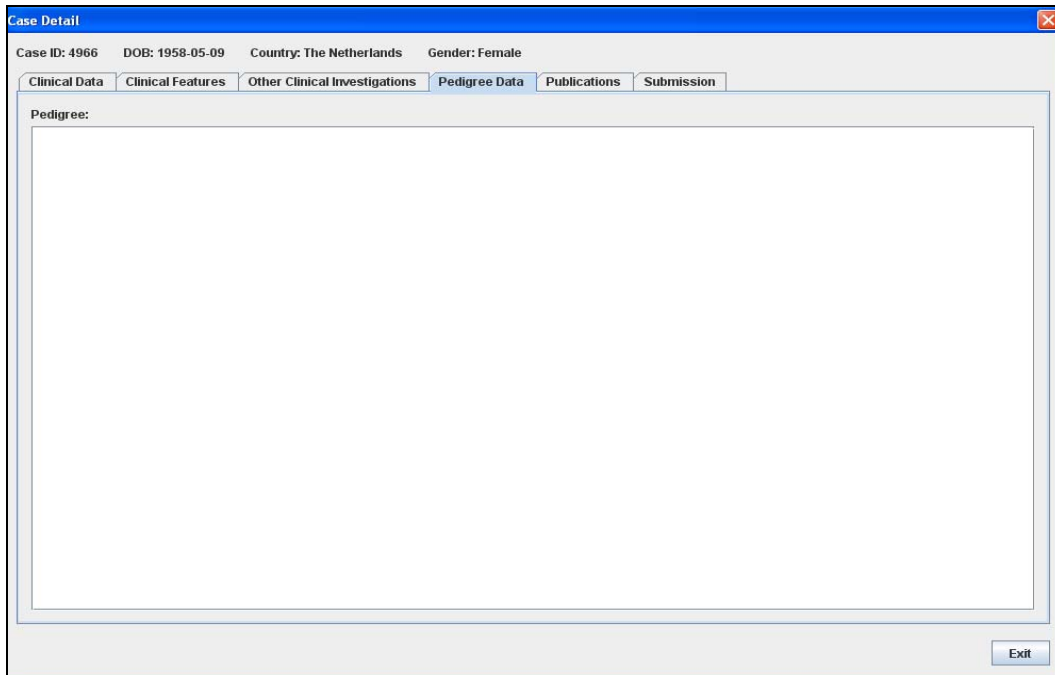
Buttons for "OK" and "Cancel" are at the bottom of the dialog. In the background, buttons for "Add", "Modify", "Delete", and "Exit" are visible at the bottom of the main window.

Choose a type of investigation from the Standard Investigation Test list or enter a short description in the field Other Investigation Test.

Enter the test result and the date of testing and press the button “OK”. The entry is now displayed on the “Other Clinical Investigations” tab.

If you either want to edit or remove an existing entry, highlight the chosen entry and press the button “Modify” or “Delete” respectively.

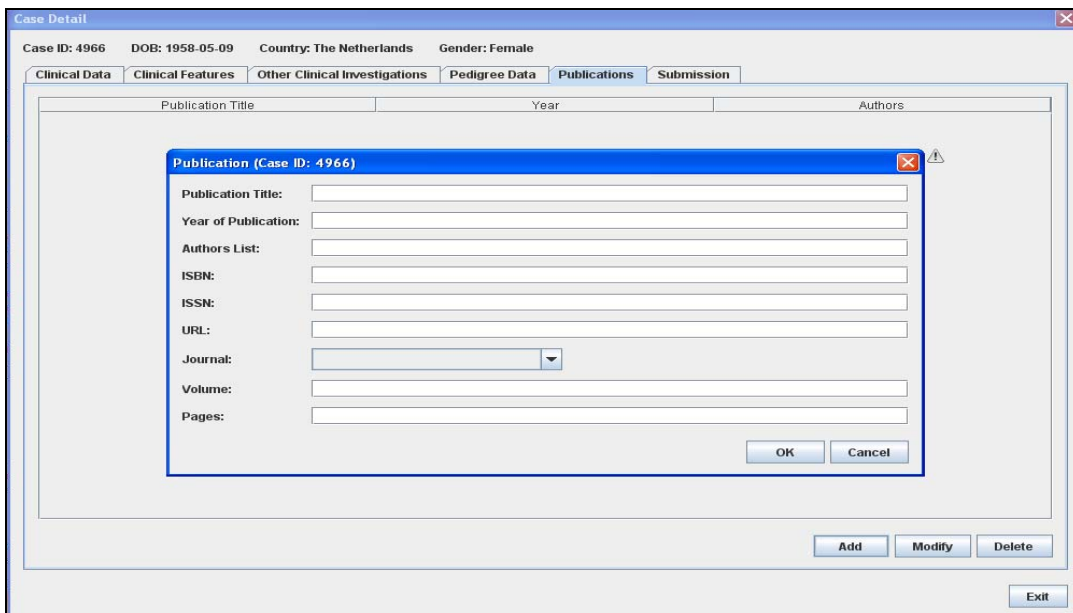
7. Enter pedigree information



The screenshot shows a software window titled "Case Detail" with a blue header bar. Below the header, the following information is displayed: Case ID: 4966, DOB: 1958-05-09, Country: The Netherlands, Gender: Female. There are six tabs: "Clinical Data", "Clinical Features", "Other Clinical Investigations", "Pedigree Data", "Publications", and "Submission". The "Pedigree Data" tab is selected, and it contains a large, empty rectangular text area for entering pedigree information. An "Exit" button is located in the bottom right corner of the window.

Pedigree data can be entered in the free text field.

8. Enter publication if this case was published



The screenshot shows the same "Case Detail" window, but now the "Publications" tab is selected. The main area contains a table with columns for "Publication Title", "Year", and "Authors". A dialog box titled "Publication (Case ID: 4966)" is open in the foreground. This dialog box contains the following fields: "Publication Title:", "Year of Publication:", "Authors List:", "ISBN:", "ISSN:", "URL:", "Journal:" (with a dropdown arrow), "Volume:", and "Pages:". At the bottom of the dialog box are "OK" and "Cancel" buttons. In the background, the "Publications" tab also shows "Add", "Modify", and "Delete" buttons at the bottom, and an "Exit" button in the bottom right corner.

Click on the tab "Publications" if you want to add a publication on the current case. Press the button "Add", the window "Publication" is displayed.

9. Submission of the case

The screenshot shows a web browser window titled "Case Detail". At the top, it displays patient information: Case ID: 4966, DOB: 1958-05-09, Country: The Netherlands, Gender: Female. Below this is a navigation bar with tabs for "Clinical Data", "Clinical Features", "Other Clinical Investigations", "Pedigree Data", "Publications", and "Submission". The "Submission" tab is active. The main content area contains the following text:

Please note that once you have registered (submitted) this case, you will not be able to edit these details or register any additional information through this web interface. If you wish to modify the submitted information please email these directly to: info@ecaruca.net

Please tick the box below to indicate that you understand this instruction and wish to submit the case details that you have registered.

After clicking the submit button the case will be removed from your list of cases that are available for registration. The submitted data will be checked by the daily management team. After approval the case will be available for account holders. An overview of the data will be sent to you by email, including the parent account details (see "parent account" in the menu bar).

Below the text is a checkbox labeled "I understand this instruction." which is checked with a red checkmark. To the right of the checkbox is a "Submit" button, which is circled in red. At the bottom right of the form is an "Exit" button.

When you have finished entering the information about the patient, click the tab "Submission".

Read the instruction and tick the box below it to indicate that you understand the procedure. Press the "Submit" button.

The case will be removed from your list of cases that are available for registration. The submitted data will be checked by the daily management team. After approval the case will be available for account holders. An overview of the data will be sent to you by email, including parent account details.

The ECARUCA parent account gives the parents access to a restricted ECARUCA webpage that includes an overview of their child's clinical data. In this way the parents can see whether the information on their child is correct. It also allows the parents to submit additional information.