

## Manual for searching the ECARUCA Database

First, log in with your account details. If you don't have an account yet, you can fill in the account request form at the log-in page.

ECARUCA

Home > Log in

**Log in**

Account name

Password

Password forgotten? Please [contact us](#) .

Not an ECARUCA-member yet? Please go to the [account request form](#) .

features, but NOT the patient material used for the analysis; this stays in the centre where the research was carried out

You will access the restricted pages of ECARUCA

Log out Help

Restricted homepage

Welcome to the restricted pages of the ECARUCA web site

★ 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](#)
- [Patient of the Month](#)

★ 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](#)

### Searching by aberration:

1. Press the link 'search by chromosome aberration' .

2. You can search on the specific aberration you are interested in. Please note, the basepair positions within the ECARUCA database are based on the UCSC Genome Browser 2004.

**Search by chromosome aberration**

Give an overview of all chromosome aberrations known to the register with a:

- deletion (del)
- duplication (dup)
- trip (trp)
- quadruplication (qdp)
- triploid
- tetraploid
- UPD
- homozygous loss
- of chromosome number
- including the region with  
breakpoint closest to pter   
or enter a start basepair   
and  
breakpoint closest to qter   
or enter an end basepair
- all cases having mosaic aberrations.
- all cases with complex rearrangements.
- all cases without microarray data.
- show only ring chromosomes

You may choose if you wish to include or exclude cases having mosaic aberrations, complex rearrangements or cases without microarray data. You can choose to show only ring chromosomes. Press 'search' to start.

The different aberrations will appear in the search screen:

**Search by chromosome aberration**

Listed are all chromosome aberrations known to the register with a DEL of chromosome [ 1 ] including the region from [ q42.2 ] to [ q44 ]:

Chromosome aberration	Chrom. No.	Cytoband	No. of aberrations	No. of aberrations part of complex rearrangement	Mark for overview of clinical features
DEL	1	<input type="text" value="q32 q42"/>	1	0	<input type="checkbox"/>
DEL	1	<input type="text" value="q41 q43"/>	1	0	<input type="checkbox"/>
DEL	1	<input type="text" value="q42 qter"/>	15	2	<input type="checkbox"/>
DEL	1	<input type="text" value="q42.3 qter"/>	8	2	<input type="checkbox"/>
DEL	1	<input type="text" value="q43 q44"/>	3	0	<input type="checkbox"/>
DEL	1	<input type="text" value="q43 qter"/>	18	8	<input type="checkbox"/>
DEL	1	<input type="text" value="q44 q44"/>	1	0	<input type="checkbox"/>
DEL	1	<input type="text" value="q44 qter"/>	13	12	<input type="checkbox"/>

For references please go to [publications](#) of registered cases with the above mentioned chromosome aberrations.

3. Press the cytoband region in which you are interested to see all cases with an aberration within this specific region.

[Print](#)

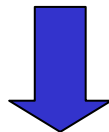
**Search by chromosome aberration**

Listed are all chromosome aberrations known to the register with a DEL of chromosome [ 1 ] including the region from [ q42.2 ] to [ q44 ]:

Chromosome aberration	Chrom. No.	Cytoband	No. of aberrations	No. of aberrations part of complex rearrangement	Mark for overview of clinical features
DEL	1	<a href="#">q32 q42</a>	1	0	<input type="checkbox"/>
DEL	1	<a href="#">q41 q43</a>	1	0	<input type="checkbox"/>
DEL	1	<a href="#">q42 qter</a>	15	2	<input type="checkbox"/>
DEL	1	<a href="#">q42.3 qter</a>	8	2	<input type="checkbox"/>
DEL	1	<a href="#">q43 q44</a>	3	0	<input type="checkbox"/>
DEL	1	<a href="#">q43 qter</a>	18	8	<input type="checkbox"/>
DEL	1	<a href="#">q44 q44</a>	1	0	<input type="checkbox"/>
DEL	1	<a href="#">q44 qter</a>	13	12	<input type="checkbox"/>

[Show overview of clinical features](#)

For references please go to [publications](#) of registered cases with the above mentioned chromosome aberrations.



[Print](#)

**Search by chromosome aberration**

The following cases were found. Please click the case of interest in order to view its details.(!Only the cases including iscn results/karyotype are displayed)

Case-id	Karyotype	Microarray data avail.
<a href="#">12</a>	46,XY,del(1)(q42.3)dn	no
<a href="#">446</a>	46,XY,del(1)(q42.3)dn	no
<a href="#">447</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">1292</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">1480</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">2270</a>	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
<a href="#">2271</a>	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
<a href="#">2485</a>	46,XY,t(1;8)(q22.1;q42.3),del(1)(q42.3)dn	no

Back to the [overview of aberrations](#)

4. Click on the specific case in which you are interested to get an overview of the clinical features of this patient.

[Print](#)

**Search by chromosome aberration**

The following cases were found. Please click the case of interest in order to view its details. (!Only the cases including iscn results/karyotype are displayed)

Case-id	Karyotype	Microarray data avail.
<a href="#">12</a>	46,XY,del(1)(q42.3)dn	no
<a href="#">446</a>	46,XY,del(1)(q42.3)dn	no
<a href="#">447</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">1292</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">1480</a>	46,XX,del(1)(q42.3)dn	no
<a href="#">2270</a>	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
<a href="#">2271</a>	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
<a href="#">2485</a>	46,XY,t(1;8)(q22.1;q42.3),del(1)(q42.3)dn	no

Back to the [overview of aberrations](#)



**Case Details:**

Case ID: 1480

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**Personal data:**

Sex: Female

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**Clinical data:**

Age at last exam: 7 month(s)

Clinical feature:

- Build
  - Low birth-weight (< 3rd centile)
- Cranium
  - Cerebral atrophy/heterotopia
  - Agensis/hypopl. of corpus callosum
  - Hydrocephaly/Large ventricles non-spec
- Hair
  - Sparse hair/alopecia areata
- Ears
  - Dysplastic ears
  - Low-set ears
- Eyes,associated structures
  - Palpebral fissures slant up
  - Epicanthic folds
- Nose
  - Depressed/flat nasal bridge
  - Broad nasal tip
- Face
  - Round face
  - Small mandible/micrognathia
- Mouth
  - Down-turned corners of the mouth
  - Cupid bow shape of mouth
  - Simple/Absent philtrum
  - Thin upper lip
- Oral region
  - Cleft palate
- Voice
  - High pitched voice
- Thorax
  - Atrial septum defect
  - Pulmonary stenosis/atresia
- Hands
  - Small hands
  - Single palmar crease
- Feet
  - Small feet
- Neurology
  - Hypotonia
  - Mental retardation
  - Seizures/Abnormal EEG
- Gestation/delivery.
  - Gestation,37-42 weeks.
- Cranium
  - Microcephaly,prenatal onset

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**Cytogenetic data:**

Combined karyotype (ISCN 2005): 46,XX,del(1)(q42.3)dn

Aberrations:

- Aberration type: deletion (del)
- Chromosome number: 1
- Breakpoint1: q42.3
- Breakpoint2: qter
- Remark1: de novo

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**Material:** Blood Lymphocytes

**Routine chromosome analysis:** Routine GTG-banding

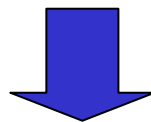
5. If you would like to view an overview off all clinical features in the different cases with a specific aberration you can mark the boxes for overview of clinical features and press the 'show overview of clinical features' button:

**Search by chromosome aberration**

Listed are all chromosome aberrations known to the register with a DEL of chromosome [ 1 ] including the region from [ q42.2 ] to [ q44 ]:

Chromosome aberration	Chrom. No.	Cytoband	No. of aberrations	No. of aberrations part of complex rearrangement	Mark for overview of clinical features
DEL	1	q32 q42	1	0	<input type="checkbox"/>
DEL	1	q41 q43	1	0	<input type="checkbox"/>
DEL	1	q42 qter	15	2	<input type="checkbox"/>
DEL	1	q42.3 qter	8	2	<input type="checkbox"/>
DEL	1	q43 q44	3	0	<input type="checkbox"/>
DEL	1	q43 qter	18	8	<input type="checkbox"/>
DEL	1	q44 q44	1	0	<input type="checkbox"/>
DEL	1	q44 qter	13	12	<input type="checkbox"/>

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**Clinical feature summary**

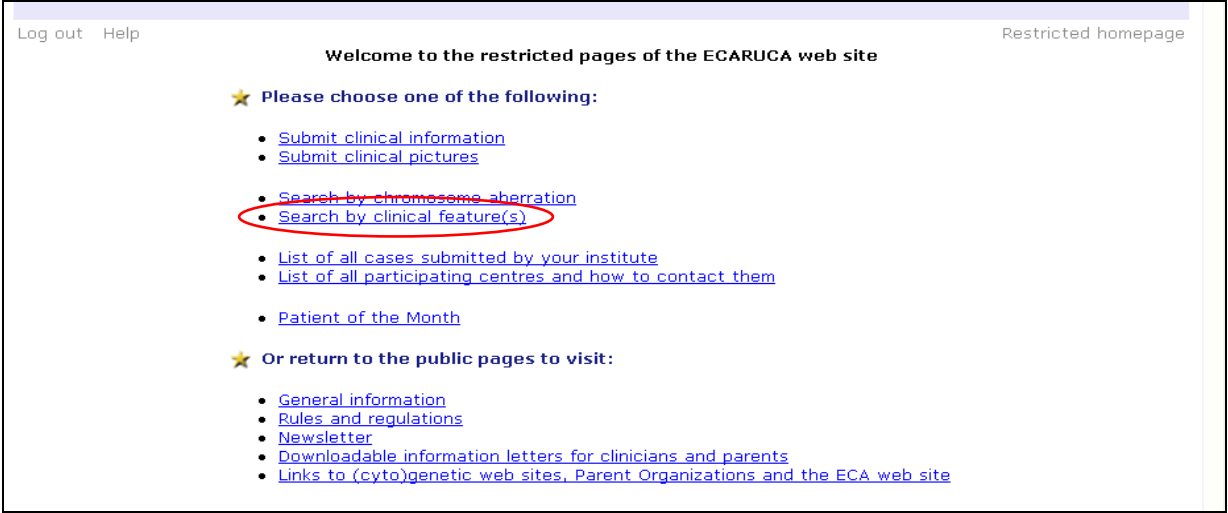
[Major anomalies](#)

[Minor anomalies](#)

**Major anomalies**

		q44-qter	q43-qter	Totals
Cranium	Agenesis/hypopl. of corpus callosum	(1/12)	(3/17)	(4/29)
Cranium	Anterior encephalocele/meningocele	(0/12)	(2/17)	(2/29)
Cranium	Posterior encephalocele	(0/12)	(1/17)	(1/29)
Ears	Pre-auricular tags	(1/12)	(0/17)	(1/29)
Ears	Narrow/Atretic auditory canal	(0/12)	(1/17)	(1/29)
Eyes, globes	Microphthalmia	(1/12)	(0/17)	(1/29)
Oral region	Cleft palate	(0/12)	(4/17)	(4/29)
Oral region	Submucous cleft palate	(0/12)	(1/17)	(1/29)
Oral region	Absent or hypoplastic tongue	(1/12)	(0/17)	(1/29)
Back and spine	Meningocele/Meningo-myelocele	(0/12)	(2/17)	(2/29)
Thorax	Aortic stenosis	(1/12)	(0/17)	(1/29)
Thorax	Coarctation of the aorta	(0/12)	(1/17)	(1/29)
Thorax	Congenital cardiac anomaly, general	(1/12)	(1/17)	(2/29)
Thorax	Patent ductus arteriosus	(0/12)	(1/17)	(1/29)
Thorax	Pulmonary stenosis/atresia	(0/12)	(1/17)	(1/29)
Thorax	Ventricular septal	(1/12)	(1/17)	(2/29)

## Searching by clinical feature:

1. After gaining access to the restricted pages of ECARUCA you can click on the link 'search by clinical feature(s)'.  


Log out Help Restricted homepage

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- [Links to \(cyto\)genetic web sites, Parent Organizations and the ECA web site](#)

**Search by clinical features**

**Standard Features for Selection:**

- ☐ Cranium,general
  - ☐ Acro/turricephaly
  - ☐ Brachycephaly
  - ☐ Dolichocephaly/Scaphocephaly
  - ☐ Macrocephaly
  - ☐ Microcephaly
  - ☐ Plagiocephaly/Asymmetrical skull
  - ☐ Platybasia
  - ☐ Trigonocephaly
  - ☐ Microcephaly,prenatal onset
  - ☐ Microcephaly,post-natal onset.
  - ☐ Macrocephaly,prenatal onset.
  - ☐ Macrocephaly,post-natal onset.
- ☐ Cranial bones,general
- ☐ Brain abnormalities,general

**Selected Features:**

- ☑ Absent fingers or oligodactyl(\*)
- ☑ Acro/turricephaly

Select Find Collapse Remove

The minimal number of features that has to be reported in the chromosome aberration:

**Include** all cases without microarray data.  
**Exclude**

**Include** all cases having mosaic aberrations.  
**Exclude**

**Include** all cases with complex rearrangements.  
**Exclude**

show only ring chromosomes.

**Search**

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.

2. 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:

1. expand the tree by double-clicking on the category (and sub-categories if applicable) until the desired feature is listed.
2. search for a feature by pressing the “find” button, the Feature Search window is displayed.

When you have found the clinical feature you wish to include in your search, add it to the case by clicking this feature and then the select button. The selected features will be visible in the Selected Features tabe. If you wish to remove features, select the specific feature and click the ‘remove’ button.

**Search by clinical features**

**Standard Features for Selection:**

- Cranium,general
  - Acro/turricephaly
  - Brachycephaly
  - Dolichocephaly/Scaphocephaly
  - Macrocephaly
  - Microcephaly
  - Plagiocephaly/Asymmetrical skull
  - Platybasia
  - Trigonocephaly
  - Microcephaly,prenatal onset
  - Microcephaly,post-natal onset.
  - Macrocephaly,prenatal onset.
  - Macrocephaly,post-natal onset.
- Cranial bones,general
- Brain abnormalities,general

**Selected Features:**

- Absent fingers or oligodactyl(\*)
- Acro/turricephaly

**Select** **Find** **Collapse** **Remove**

The minimal number of features that has to be reported in the chromosome aberration:

**Include** **Exclude** all cases without microarray data.  
**Include** **Exclude** all cases having mosaic aberrations.  
**Include** **Exclude** all cases with complex rearrangements.

show only ring chromosomes.

**Search**

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.

You may choose if you wish to include or exclude cases having mosaic aberrations, complex rearrangements or cases without microarray data. You can select to show only ring chromosomes. Press ‘search’ to start.

The different aberrations will appear in the search screen.

**Search by chromosome aberration**

The following aberrations contain cases having the features that you sought for:

<b>Chromosome aberration</b>	<b>Chrom. No.</b>	<b>Cytoband</b>	<b>No. of aberrations</b>	<b>No. of aberrations part of complex rearrangement</b>	<b>Mark for overview of clinical features</b>
DEL	13	<input type="text" value="q22 qter"/>	1	0	<input type="checkbox"/>
DEL	13	<input type="text" value="q32 qter"/>	1	0	<input type="checkbox"/>

For references please go to [publications](#) of registered cases with the above mentioned chromosome aberrations.

See further point 3 in the section "Searching by aberration".