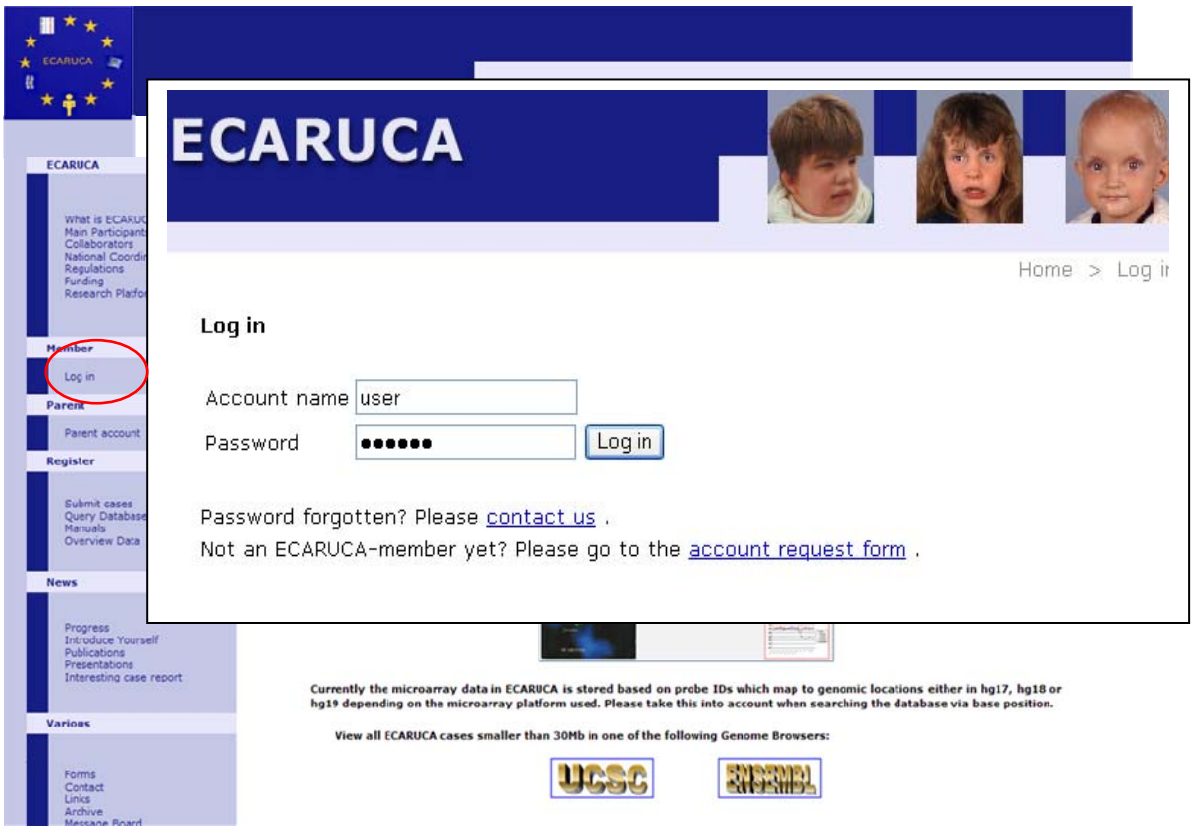
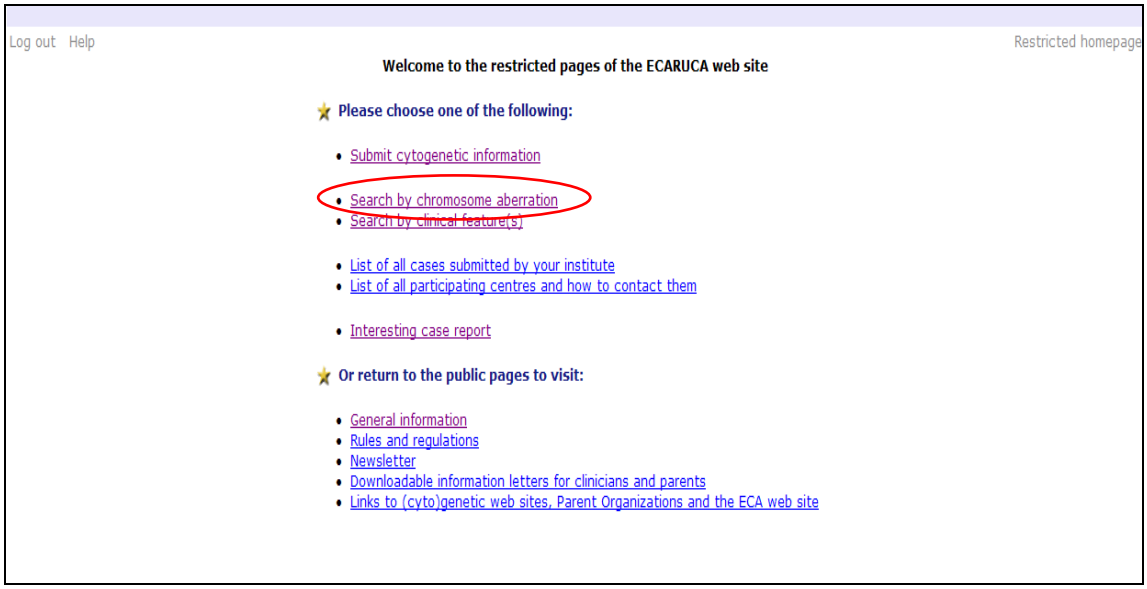
Manual for searching the ECARUCA Database

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.



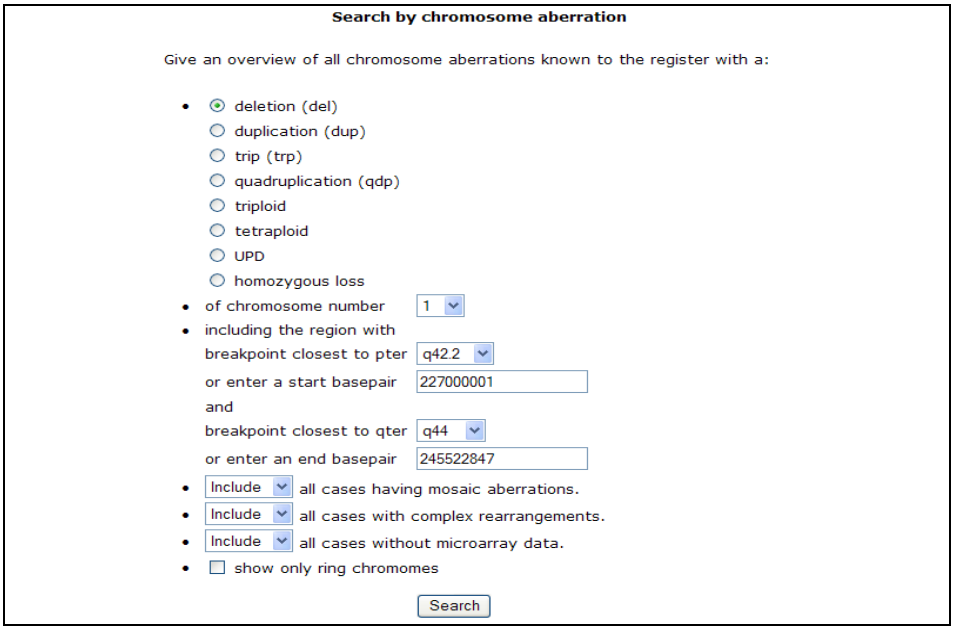
You will access the restricted pages of ECARUCA



Searching by aberration:

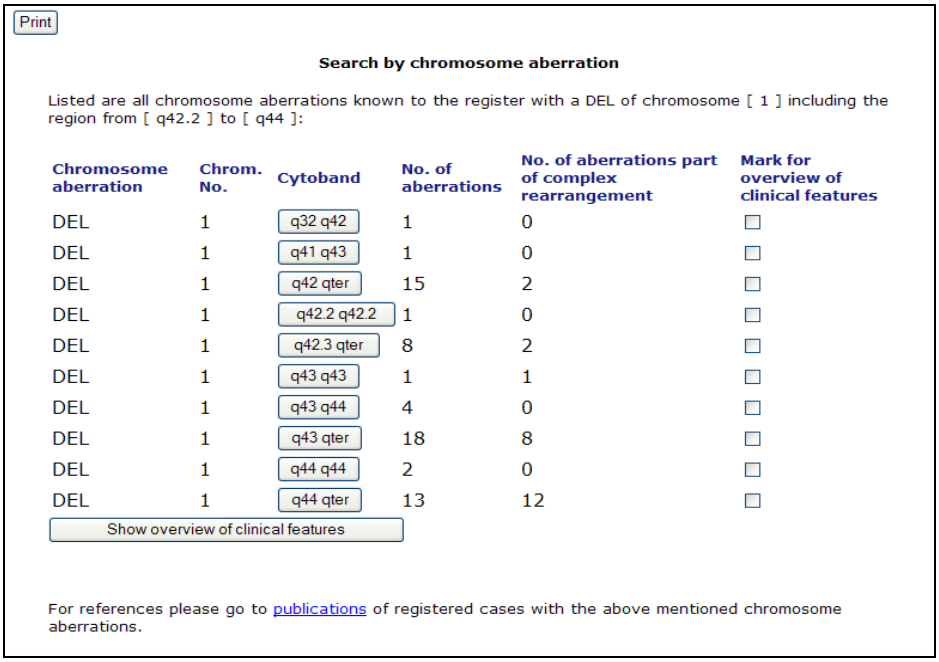
1. Press the link ‘search by chromosome aberration’ .

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.

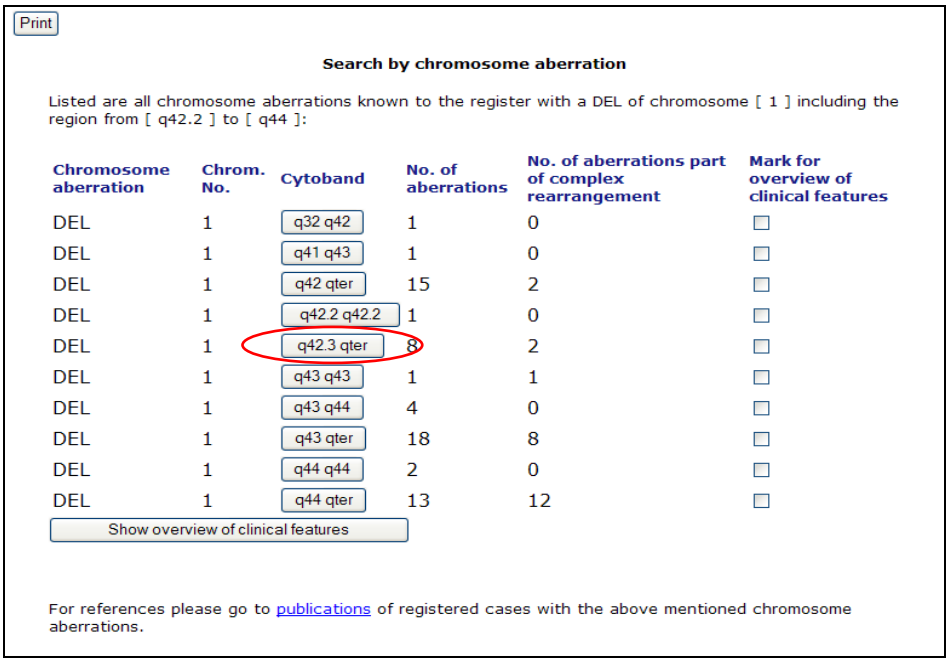


You may choose if you wish to include or exclude cases having mosaic aberrations, complex rearrangements or cases without microarray data. You can choose to show only ring chromosomes. Press ‘search’ to start.

The different aberrations will appear in the search screen:



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



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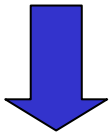
Search by chromosome aberration

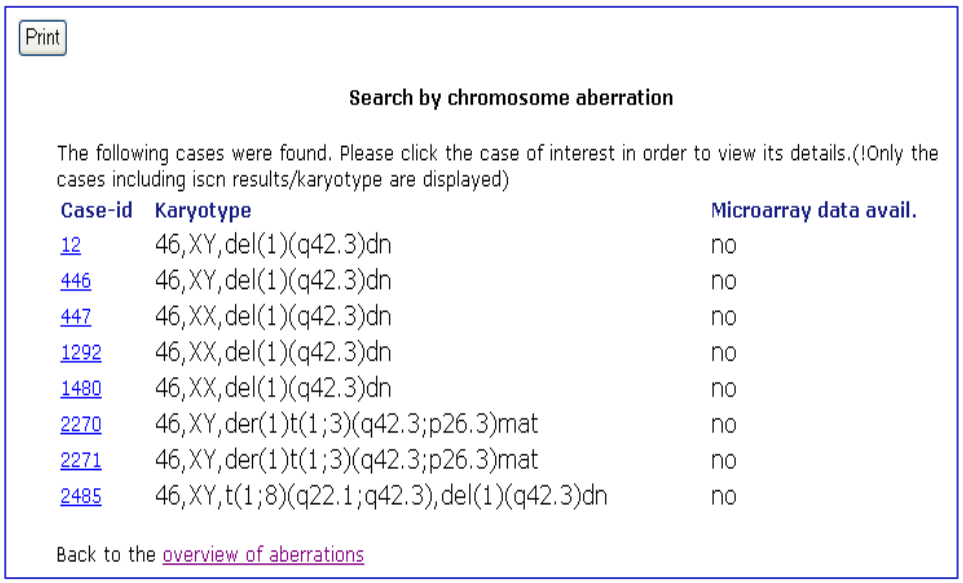
Listed are all chromosome aberrations known to the register with a DEL of chromosome [1] including the region from [q42.2] to [q44]:

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	☐
DEL	1	q41 q43	1	0	☐
DEL	1	q42 qter	15	2	☐
DEL	1	q42.2 q42.2	1	0	☐
DEL	1	q42.3 qter	8	2	☐
DEL	1	q43 q43	1	1	☐
DEL	1	q43 q44	4	0	☐
DEL	1	q43 qter	18	8	☐
DEL	1	q44 q44	2	0	☐
DEL	1	q44 qter	13	12	☐

Show overview of clinical features

For references please go to publications of registered cases with the above mentioned chromosome aberrations.





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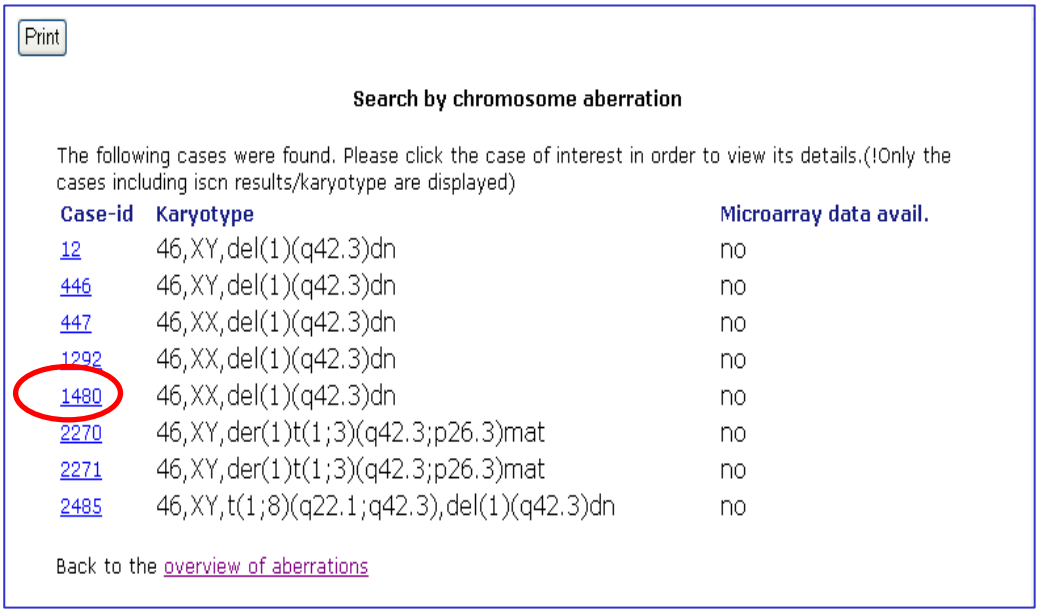
Search by chromosome aberration

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.
12	46,XY,del(1)(q42.3)dn	no
446	46,XY,del(1)(q42.3)dn	no
447	46,XX,del(1)(q42.3)dn	no
1292	46,XX,del(1)(q42.3)dn	no
1480	46,XX,del(1)(q42.3)dn	no
2270	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
2271	46,XY,der(1)t(1;3)(q42.3;p26.3)mat	no
2485	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.



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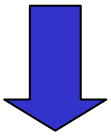
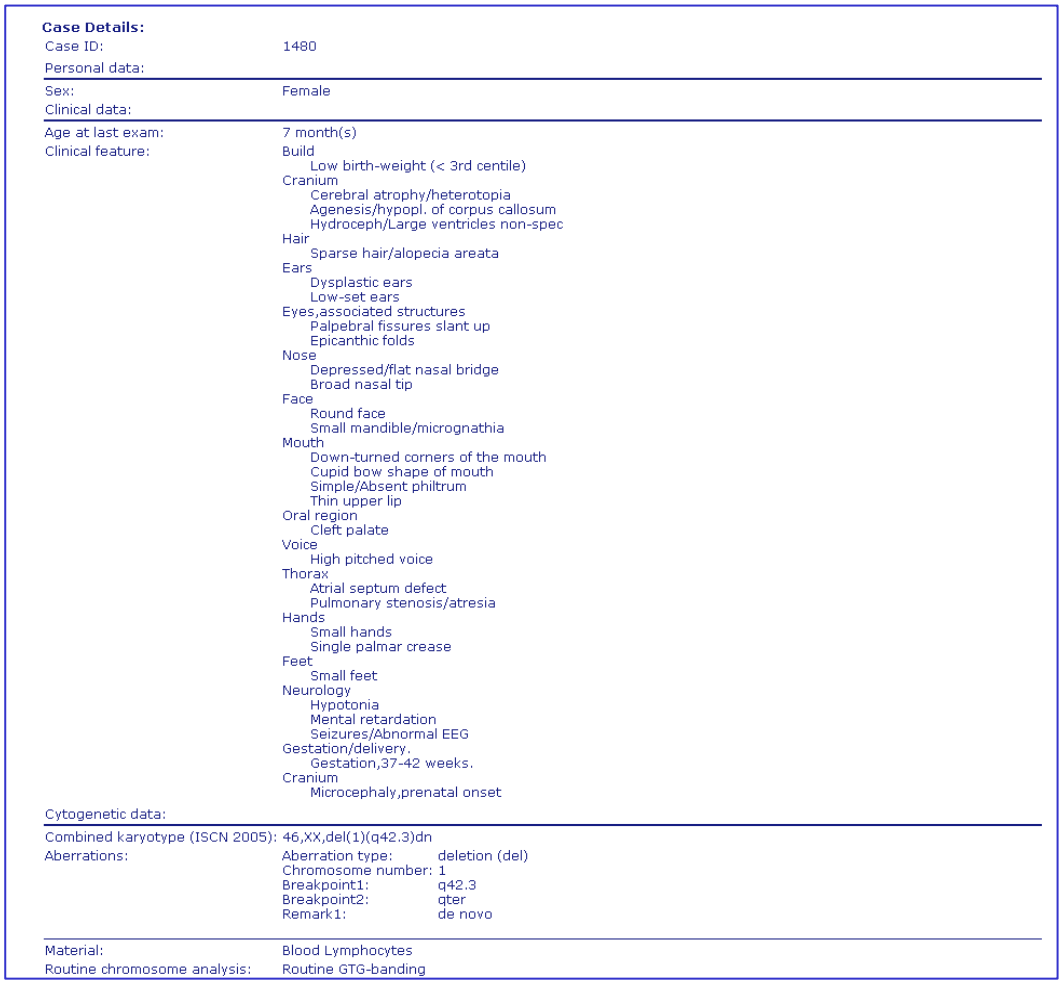
Back to the overview of aberrations





Case Details:

Case ID: 1480

Personal data:

Sex: Female

Clinical data:

Age at last exam: 7 month(s)

Clinical feature:

Build
- Low birth-weight (< 3rd centile)

Cranium
- Cerebral atrophy/heterotopia
- Agenesis/hypopl. of corpus callosum
- Hydroceph/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

Cytogenetic data:

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

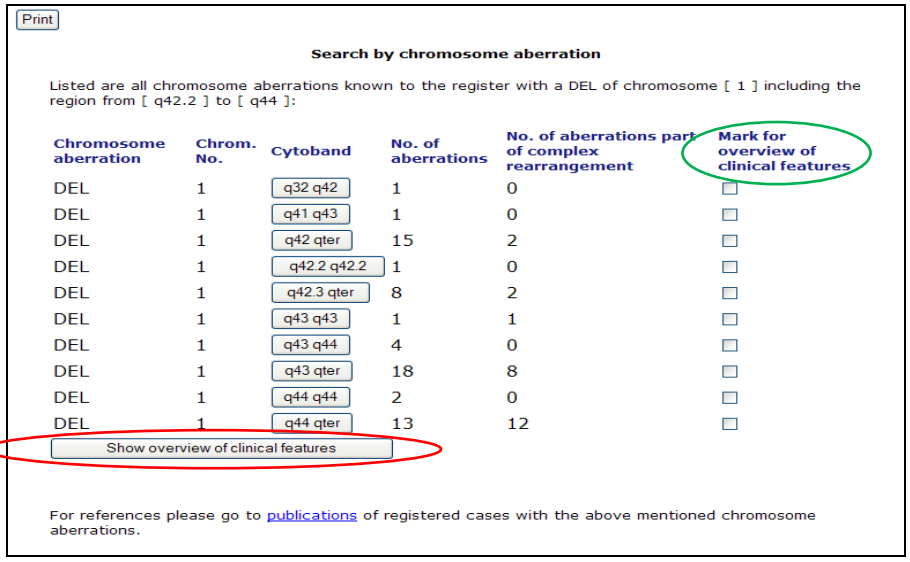
Aberrations:

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

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:



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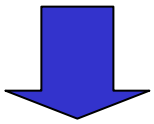
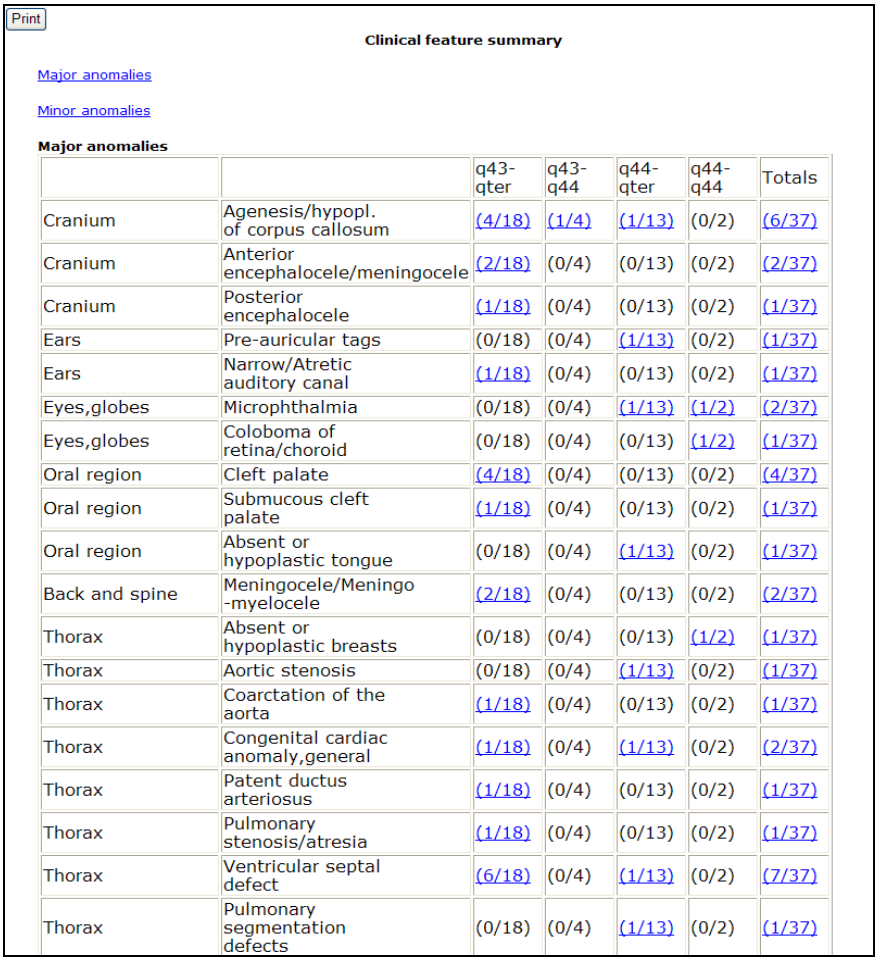
DEL	1	q43 q44	4	0	☐
DEL	1	q43 qter	18	8	☐
DEL	1	q44 q44	2	0	☐
DEL	1	q44 qter	13	12	☐

Show overview of clinical features

For references please go to publications of registered cases with the above mentioned chromosome aberrations.





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Clinical feature summary

Major anomalies

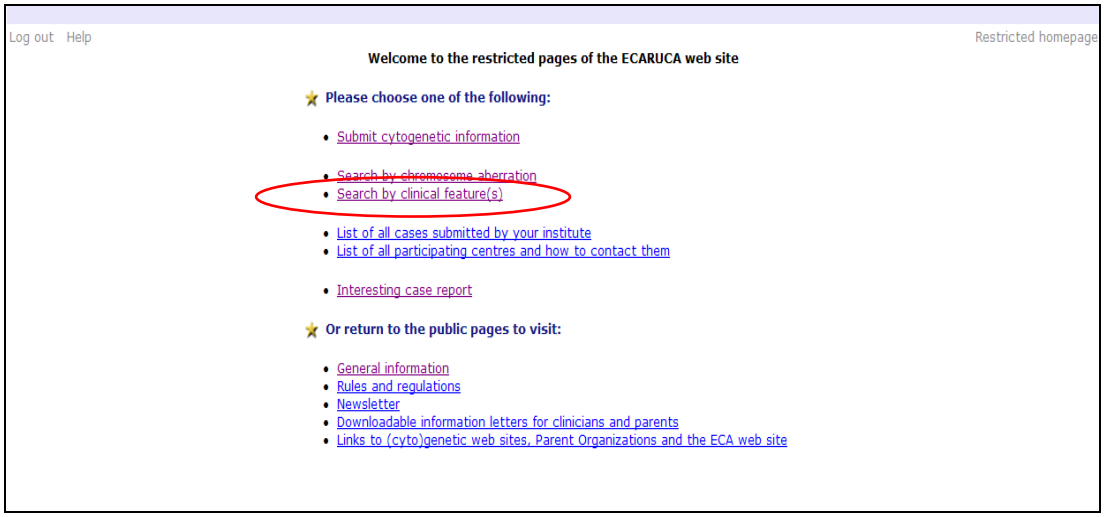
Minor anomalies

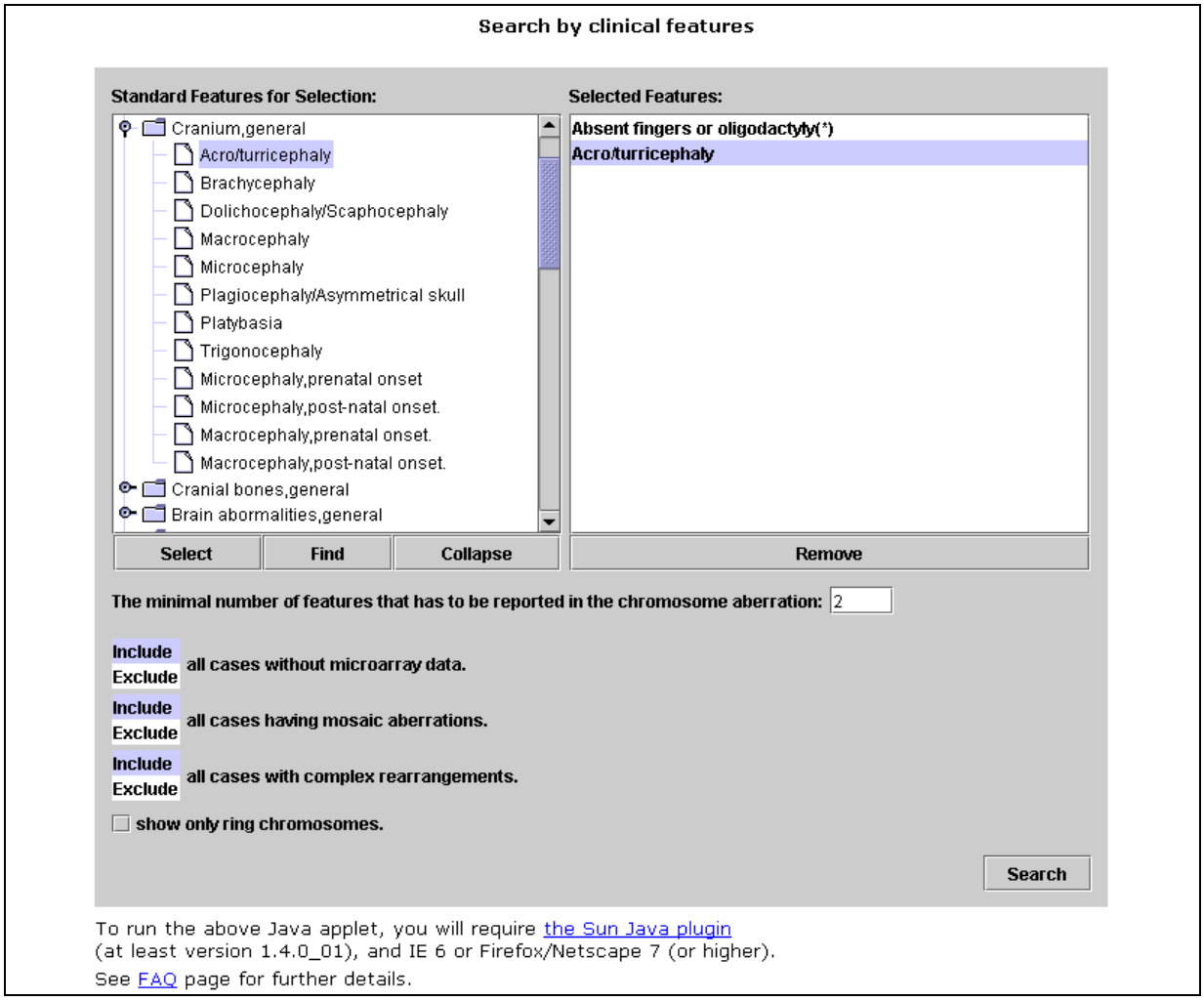
Major anomalies

		q43-qter	q43-q44	q44-qter	q44-q44	Totals
Cranium	Agenesis/hypopl. of corpus callosum	(4/18)	(1/4)	(1/13)	(0/2)	(6/37)
Cranium	Anterior encephalocele/meningocele	(2/18)	(0/4)	(0/13)	(0/2)	(2/37)
Cranium	Posterior encephalocele	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Ears	Pre-auricular tags	(0/18)	(0/4)	(1/13)	(0/2)	(1/37)
Ears	Narrow/Atretic auditory canal	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Eyes,globes	Microphthalmia	(0/18)	(0/4)	(1/13)	(1/2)	(2/37)
Eyes,globes	Coloboma of retina/choroid	(0/18)	(0/4)	(0/13)	(1/2)	(1/37)
Oral region	Cleft palate	(4/18)	(0/4)	(0/13)	(0/2)	(4/37)
Oral region	Submucous cleft palate	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Oral region	Absent or hypoplastic tongue	(0/18)	(0/4)	(1/13)	(0/2)	(1/37)
Back and spine	Meningocele/Meningo -myelocele	(2/18)	(0/4)	(0/13)	(0/2)	(2/37)
Thorax	Absent or hypoplastic breasts	(0/18)	(0/4)	(0/13)	(1/2)	(1/37)
Thorax	Aortic stenosis	(0/18)	(0/4)	(1/13)	(0/2)	(1/37)
Thorax	Coarctation of the aorta	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Thorax	Congenital cardiac anomaly,general	(1/18)	(0/4)	(1/13)	(0/2)	(2/37)
Thorax	Patent ductus arteriosus	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Thorax	Pulmonary stenosis/atresia	(1/18)	(0/4)	(0/13)	(0/2)	(1/37)
Thorax	Ventricular septal defect	(6/18)	(0/4)	(1/13)	(0/2)	(7/37)
Thorax	Pulmonary segmentation defects	(0/18)	(0/4)	(1/13)	(0/2)	(1/37)

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).



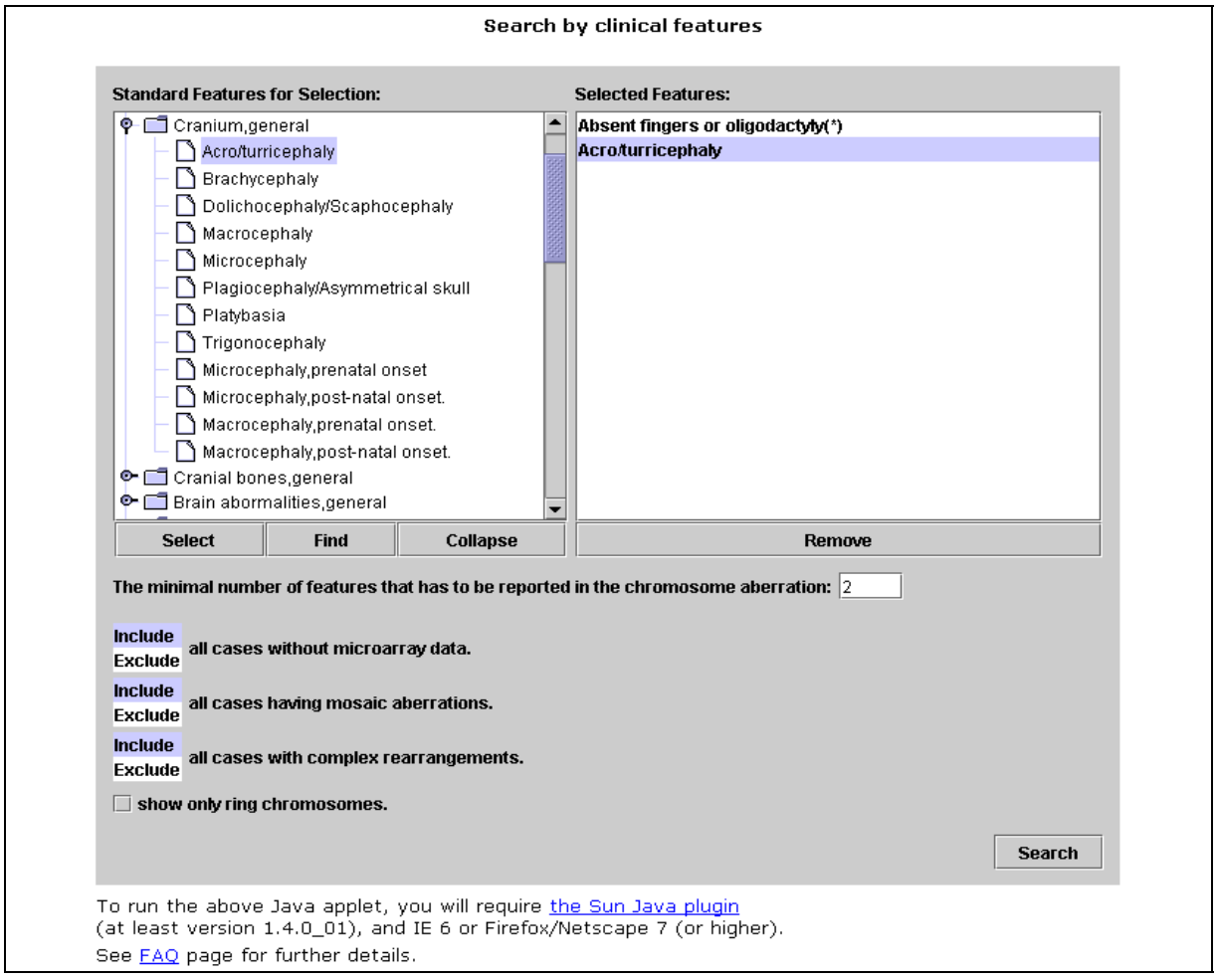


2. In the section Standard Features, an expandable tree of clinical features is shown. The tree of features is a hierarchical structure in which the features are divided into (sub)categories. A description of a feature is obtained by right-clicking on the feature of choice.

There are two ways of finding a specific clinical feature:

- 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 the clinical feature you wish to include in your search, add it to the case by clicking this feature and than 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.



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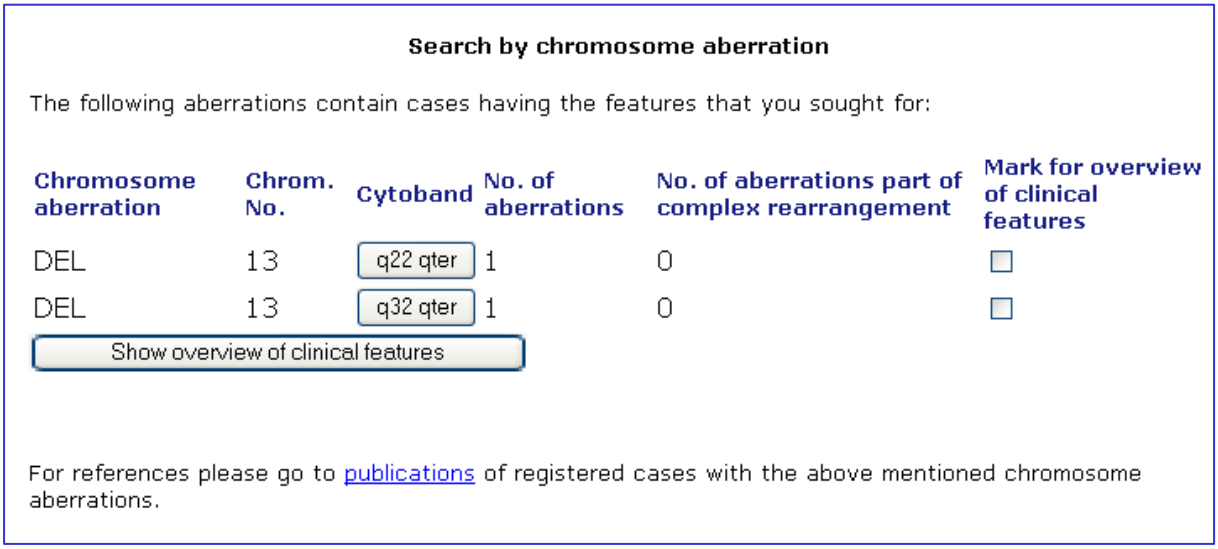
The different aberrations will appear in the search screen.



See further point 3 in the section “Searching by aberration”.