



CANCER GENOMICS CONSORTIUM

Educating for Best Practices in Clinical Cancer Genomics

Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer

Mitelman Database of Chromosome
Aberrations and Gene Fusions in Cancer
(2017). Mitelman F, Johansson B and Mertens
F (Eds.),

[http://cgap.nci.nih.gov/Chromosomes/Mitel
man](http://cgap.nci.nih.gov/Chromosomes/Mitelman)

CANCER GENOME ANATOMY PROJECT

CGAP How To

Genes

Chromosomes

Tissues

SAGE Genie

RNAi

Pathways

Tools



Chromosomes

Tools

- FISH-mapped BACs
- Genetic and Physical SNP Maps
- Mitelman Searchers
- SNP500Cancer

CGAP Data

- Download

Purchase CGAP Reagents

- CCAP BAC Clones

Related Links

- Atlas of Genetics & Cytogenetics in Oncology & Haematology
- Progenetix
- SKY/M-FISH & CGH Database

Quick Links:

- ICG
- NCI Home
- NCICB Home
- NCBI Home
- OCG

Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer

Searching the Database

The information in the Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer relates chromosomal aberrations to tumor characteristics, based either on individual cases or as reported by Felix Mitelman, Bertil Johansson, and Fredrik Mertens.

CGAP has developed six web search tools to help you analyze the information within the Mitelman Database:

- The **Cases Quick Searcher** allows you to query the individual patient cases using the four major fields: aberration, breakpoint, morphology, and topography.
- The **Cases Full Searcher** permits a more detailed search of the same individual patient cases as above, by including more cytogenetic field choices and adding search fields for patient characteristics and references.
- The **Molecular Biology Associations Searcher** does not search any of the individual patient cases. It searches studies pertaining to gene rearrangements as a consequence of cytogenetic aberrations.
- The **Clinical Associations Searcher** does not search any of the individual patient cases. It searches studies pertaining to clinical associations of cytogenetic aberrations and/or gene rearrangements.
- The **Recurrent Chromosome Aberrations Searcher** provides a way to search for structural and numerical abnormalities that are recurrent, i.e., present in two or more cases with the same morphology and topography.
- The **Reference Searcher** queries only the references themselves, i.e., the references from the individual cases and the molecular biology and clinical associations.

Database last updated on February 20, 2017

Total number of cases = **66,919**

Total number of gene fusions = **10,676**

Need help! To learn about the Mitelman Database and how to search it, please visit:

- All about the Mitelman Database, which provides background information about the Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer.
- Mitelman Database Search Help, which contains information on how to use the search tools.
- ISCN Abbreviated Terms and Symbols, which provides a list of terms and symbols used to describe chromosome abnormalities.

Citation of the Database

To cite the use of the Mitelman Database from this CGAP Website in a publication, please quote the following: "**Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer**"
<http://cgap.nci.nih.gov/Chromosomes/Mitelman>

The Mitelman Database is supported by the Swedish Cancer Society and the Swedish Childhood Cancer Foundation. The database is updated quarterly in February, May, August, and November.

5 main tools

Curation Activity

Online education materials

Filling Database Search Fields

- <https://cgap.nci.nih.gov/Chromosomes/Help>
- Review this page before attempting to enter information into search fields.
- Make sure ISCN nomenclature is correct when searching for a specific abnormality.
 - <https://cgap.nci.nih.gov/Chromosomes/ISCNSymbols>
 - <https://www.amazon.com/ISCN-2016-International-Cytogenomic-Nomenclature/dp/3318058572>

Cases Quick Searcher

Mitelman Cases Quick Searcher

The Quick Searcher, like the Full Searcher, analyzes the individual patient cases in the Mitelman Database. However, unlike the Full Searcher, it limits the search fields to four criteria: abnormality, breakpoint, topography, and morphology. It finds all cases that match the chosen criteria, organizes them by lead author in the reference, and provides a link to the individual patient information and to the reference itself.

Cases Quick Searcher

Brief instructions for using the Quick Searcher:

- Check "Yes" in the "Sole Abnormality" field to view cases with only one aberration, irrespective of any other item selected.
- Choose one or more of the following five fields to query the database.
- The default setting for both the Topography and Morphology fields is all items. Or select one or more items in each scroll down box.
- Click the link at Special Morphology to view a list of all special morphology terms.
- Press Submit Query, or press Reset to begin a new search.

Sole Abnormality: ☒ No ☐ Yes

Abnormality: ☒ And ☐ Or

del(5)(q22q25)

Breakpoint: ☒ And ☐ Or

Topography:

[Expand Topography List:](#)

Adrenal
Anus
Bladder
Blood vessel
Bone and soft tissues (all sites)

Morphology:

[Expand Morphology List:](#)

Acinic cell carcinoma
Acute basophilic leukemia
Acute eosinophilic leukemia
Acute erythroleukemia (FAB type M6)
Acute lymphoblastic leukemia/lymphoblastic lymphoma

Special Morphology:

or

- Abnormality : enter appropriate Cytogenetic ISCN
- Breakpoint: enter appropriate chromosome band (ex: 5q25)
- Wildcard "*" symbol can be used if complete breakpoints are not defined. For example:
 - del(5)(q22)*
 - del(5)(q2*)
 - Or designate Breakpoint as 5q2*
- Cannot use sub-bands
 - Ex/will not recognize "5q31.1", must be typed as "5q31"

Cases Quick Searcher Results

- Query for del(5)(q22q25)

Link to information on publication

Link to information on patient/case

Chromosome Aberration Case List

Content-type: text/plain
[\[Full Text\]](#)
Displaying Investigations 1 to 1 of 1 found
From 1 References and 1 Cases

[Yunis et al 1984, N Engl J Med](#)

Case No. 29

Acute myeloblastic leukemia with maturation (FAB type M2)
45,XX,del(5)(q22q25),del(7)(q22q32),hsr(8)(p23),hsr(11)(q25)x2,-18,add(22)(q13)

Information on Patient/Case

Chromosome Aberration Case Info	
Yunis et al 1984, N Engl J Med	
Case Number 29	
Karyotype	
45,XX,del(5)(q22q25),del(7)(q22q32),hsr(8)(p23),hsr(11)(q25)x2,-18,add(22)(q13)	
Patient Characteristics	
Sex	Female
Age	70
Race	
Country	United States
Series	Unselected
Hereditary Disorder	
Present Tumor	
Topography	
Immunophenotype	
Morphology	Acute myeloblastic leukemia with maturation (FAB type M2)
Tissue	Bone marrow
Previous Tumor	
Topography	
Morphology	
Treatment	

Publication Information

Chromosome Aberration Reference Info

Reference Info

Reference Number 1100

Title High-resolution chromosomes as an independent prognostic indicator in adult acute nonlymphocytic leukemia.

Authors Yunis JJ, Brunning RD, Howe RB, Lobell M,

Journal N Engl J Med

Volume 311:812-818

Year 1984

[PubMed](#) [All Cases](#) [ClinAssoc](#)

- PubMed: Links to PubMed entry for this reference
- All Cases: List of all cases from this publication
- ClinAssoc : Clinical associations based on parameters of abnormality

Publication Information – All Cases and ClinAssoc

Chromosome Aberration Case List	
Content-type: text/plain	
[Full Text]	
Displaying Investigations 1 to 33 of 33 found	
From 1 References and 33 Cases	
<hr/>	
Yunis et al 1984, N Engl J Med	
<hr/>	
Case No. 1	Acute monoblastic leukemia with differentiation (FAB type M5b) 46,XY,inv(16)(p13q22)
Case No. 10	Acute myeloblastic leukemia with maturation (FAB type M2) 47,XY,+8
Case No. 11	Acute monoblastic leukemia without differentiation (FAB type M5a) 46,XX,t(9;11)(p22;q23)
Case No. 12	Acute myeloblastic leukemia with maturation (FAB type M2) 46,XX,t(6;9)(p22;q34)
Case No. 13	Acute myeloblastic leukemia without maturation (FAB type M1) 46,XY,t(8;9;22)(p21;q34;q11)/47,idem,+21
Case No. 14	Acute myeloblastic leukemia without maturation (FAB type M1) 46,XX,t(9;22)(q34;q11)
Case No. 15	Acute monoblastic leukemia with differentiation (FAB type M5b) 46,XX,del(9)(q13q22)
Case No. 16	Acute myeloblastic leukemia with maturation (FAB type M2) 46,XX,del(21)(q22)
Case No. 17	Acute myeloblastic leukemia without maturation (FAB type M1) 45,X,-X/45,idem,t(17;18)(p11;p11)

- All Cases: Lists cases associated with the publication with karyotypes and disease classification.

- ClinAssoc: Clinical Associations affirmed by publication

Association List	
[Full Text]	
Displaying Investigations 1 to 1 of 1 found	
From 1 References	
<hr/>	
Kasyan et al 2010, Mod Pathol	
<hr/>	
Acute erythroleukemia (FAB type M6)	

Cases Full Searcher

More “advanced” version of quick searcher retrieves same information as Quick Searcher

Cases Full Searcher

Brief instructions for using the Full Searcher:

- Check “Yes” in the “Sole Abnormality” field to view cases with only one aberration, irrespective of other items selected.
- Choose any one or more individual sections (e.g., Cytogenetic Characteristics, Patient Characteristics, etc.). N.B. Check “Sole Abnormality” to view cases with only one aberration irrespective of any other item.
- Complete as many fields within an individual section as required.
- The default setting for all expanded select boxes is all items. Or select single or multiple items.
- Click the link at Special Hereditary Disorder to view a list of all special hereditary disorder terms.
- Click the link at Special Morphology to view a list of all special morphology terms.
- Press Submit Query, or press Reset to begin a new search.

Cytogenetic Characteristics

Sole Abnormality: ☒ No ☐ Yes

Abnormality: ☒ And ☐ Or

Breakpoint: ☒ And ☐ Or

Number of Clones:

Number of Chromosomes:

Patient Characteristics

Sex: ☒ Any ☐ Male ☐ Female

Age:

Race: ☐ Asian ☐ Black ☐ White ☐ Other

Country: [Expand Country List](#)

Series:

Hereditary Disorder: ☐ Autosomal dominant ☐ Autosomal recessive ☐ Chromosome abnormality ☐ Chromosome breakage syndr ☐ Mitochondrial ☐ Sex linked

[Special Hereditary Disorder](#):

Present Tumor

Topography: [Expand Topography List](#)

Immunophenotype: ☐ B Lineage ☐ T Lineage

Morphology: [Expand Morphology List](#)

Special Morphology:

Tissue:

Previous Tumor

Previous Tumor: ☒ Any ☐ Yes ☐ No/Unknown

Topography: [Expand Topography List](#)

Morphology: [Expand Morphology List](#)

Treatment:

Reference

Authors:

Journal:

Year:

Specific ID Number: Ref. No. Case No. Inv. No.

or

Molecular Biology Associations Searcher

Molecular Biology Associations Searcher

Brief instructions for using the Molecular Biology Associations Searcher:

- Select an Association.
- Complete as many of the fields following as required.
- The default setting for both Topography and Morphology is all items. Or highlight single or multiple items.
- Any section may be left blank.
- Press Submit Query, or Reset to begin a new search.

Cytogenetic Characteristics

Abnormality: ☐ And ☐ Or

Breakpoint: ☐ And ☐ Or

Gene Characteristics

Gene: ☐ And ☐ Or

Expand Gene List:

- NUP98/LNP1
- NUP98/NSD1**
- NUP98/PHF23
- NUP98/POU1F1
- NUP98/PRRX1

Tumor Characteristics

Topography:
Expand Topography List:

- Anus
- Bladder
- Blood vessel
- Bone and soft tissues (all sites)

Morphology:
Expand Morphology List:

- Acute myeloid leukemia (all subtypes)**
- Acute myeloid leukemia, NOS
- Acute myeloid leukemia, special type
- Acute myelomonocytic leukemia (FAB type M4)
- Acute panmyelosis with myelofibrosis

Immunophenotype: ☐ B Lineage ☐ T Lineage

Reference

Authors:

Journal:

Year:

Specific ID Number: Ref. No. Inv. No.

or



Association List
<p>[Full Text]</p> <p>Displaying Investigations 1 to 13 of 13 found From 9 References</p>
<p>Brown et al 2002, Blood</p> <p>Acute myeloblastic leukemia without maturation (FAB type M1) NUP98/NSD1 t(5;11)(q35;p15)</p> <p>Acute myeloblastic leukemia with maturation (FAB type M2) NUP98/NSD1 t(5;11)(q35;p15)</p>
<p>Cui et al 2016, Leuk Lymphoma</p> <p>Acute myeloid leukemia, NOS NUP98/NSD1 t(5;11)(q35;p15)</p>
<p>Fasan et al 2013, Leukemia</p> <p>Acute myeloblastic leukemia without maturation (FAB type M1) NUP98/NSD1 t(5;11)(q35;p15)</p> <p>Acute myelomonocytic leukemia (FAB type M4) NUP98/NSD1 t(5;11)(q35;p15)</p>
<p>Hollink et al 2011, Blood</p> <p>Acute myelomonocytic leukemia (FAB type M4) NUP98/NSD1 t(5;11)(q35;p15)</p> <p>Acute monoblastic leukemia (FAB type M5) NUP98/NSD1 t(5;11)(q35;p15)</p>

Association List

[Full Text]

Displaying Investigations 1 to 13 of 13 found
From 9 References

Brown et al 2002, Blood

Acute myeloblastic leukemia without
maturation (FAB type M1)

NUP98/NSD1

t(5;11)(q35;p15)

Acute myeloblastic leukemia with
maturation (FAB type M2)

NUP98/NSD1

t(5;11)(q35;p15)

Cui et al 2016, Leuk Lymphoma

Acute myeloid leukemia, NOS

NUP98/NSD1

t(5;11)(q35;p15)

Fasan et al 2013, Leukemia

Acute myeloblastic leukemia without
maturation (FAB type M1)

NUP98/NSD1

t(5;11)(q35;p15)

Acute myelomonocytic leukemia (FAB type
M4)

NUP98/NSD1

t(5;11)(q35;p15)

Hollink et al 2011, Blood

Acute myelomonocytic leukemia (FAB type
M4)

NUP98/NSD1

t(5;11)(q35;p15)

Acute monoblastic leukemia (FAB type M5)

NUP98/NSD1

t(5;11)(q35;p15)

- For each Association on the resulting list there are links to
 - Publication Information (See slide 7)
 - Gene Information

Gene Information Page

- Click on gene name to retrieve gene information
 - Database Links, sequence IDs, Cytogenetic Location, UCSC gene/exon info
 - Links to expression visualization
 - SAGE Anatomic Viewer, Digital Northern, and 2D array displays
 - MGC Clones (NIH, NCBI)
 - Protein information
 - Homology
 - Gene Ontology

Recurrent Chromosome Aberrations Searcher

- Search for structural or numerical cytogenetic aberrations

Structural aberrations:
☐
Breakpoint:

Aberration type:

All
Balanced
Unbalanced

Numerical aberrations:
☒
Chromosome:

Aberration type:

All
Trisomy
Monosomy

Topography:
[Expand Topography List:](#)

All topographies
Adrenal
Anus
Bladder
Blood vessel

Morphology:
[Expand Morphology List:](#)

All morphologies
Acinic cell carcinoma
Acute basophilic leukemia
Acute eosinophilic leukemia
Acute erythroleukemia (FAB type M6)

Gene:
[Expand Gene List:](#)

All genes
ABL1
ACBD3
ACSL6

Aberrations Searcher Results

Numerical Chromosomal Trisomy Abnormalities

Abnormality	Morphology	Topography	Cases	Genes
+5	Transitional cell carcinoma	Bladder	2	

Numerical Chromosomal Monosomy Abnormalities

Abnormality	Morphology	Topography	Cases	Genes
-5	Transitional cell carcinoma	Bladder	13	

- Links to Morphology listed in table, Topography listed in table, and Cases

Morphology/Topography

Balanced Chromosomal Abnormalities					
Band	Abnormality	Morphology	Topography	Cases	Genes
3p14	t(3;5) (p14;p14)	Transitional cell carcinoma	Bladder	2	
5p14	t(3;5) (p14;p14)	Transitional cell carcinoma	Bladder	2	
Unbalanced Chromosomal Abnormalities					
Band	Abnormality	Morphology	Topography	Cases	Genes
1p11	del(1)(p11)	Transitional cell carcinoma	Bladder	2	
1p22	del(1)(p22)	Transitional cell carcinoma	Bladder	4	
1q10	i(1)(q10)	Transitional cell carcinoma	Bladder	3	
1q12	del(1)(q12)	Transitional cell carcinoma	Bladder	2	
1q21	del(1)(q21)	Transitional cell carcinoma	Bladder	3	
3q21	add(3)(q21)	Transitional cell carcinoma	Bladder	2	
3q21	del(3)(q21)	Transitional cell carcinoma	Bladder	2	
3q25	del(3)(q25)	Transitional cell carcinoma	Bladder	2	
5p10	i(5)(p10)	Transitional cell carcinoma	Bladder	11	
5q11	del(5)(q11q13)	Transitional cell carcinoma	Bladder	2	

- If you select the morphology or topography fields from the Aberrations Searcher, you are brought to a page with 4 tables of all cytogenetic abnormalities documented under that morphology or topography.
 - Balanced Chromosomal Abnormalities
 - Unbalanced Chromosomal Abnormalities
 - Trisomy Abnormalities
 - Monosomy Abnormalities

Cases from Aberrations Searcher Results

Fadl-Elmula et al 2000, Genes Chromosomes Cancer		
Case No. 1	Transitional cell carcinoma	Bladder
62-71,XXY,+Y,+del(1)(p21),+del(2)(p13),i(5)(p10),+6,del(6)(q21)x2,-9,-9,del(10)(p12),del(14)(q24),-15,-15,del(16)(p12),-17,+18,+del(20)(p11),+21,+22,+der(?)t(?)t(?)p13)		
Fadl-Elmula et al 2001, Int J Cancer		
Case No. 19	Transitional cell carcinoma	Bladder
67-76,XXY,del(1)(q21),der(1)t(1;8)(p?q)t(1;9)(q?q),+der(1;9)(p10;p10),der(2)t(2;13)(q13;q12)t(10;13)(q11;q22)x2,+der(2)t(2;11)(q11;q13)t(1;11)(q11;q23)x2,der(2;3)(q10;q10),del(3),+der(3)t(1;3)(p?q)t(4)(p14),der(5;12)(p10;p10),+i(5)(p10)x2,del(6)(p12),+del(6)(q13),+der(6)t(4;6)(p17;q17)x2,+der(7)t(5;7)(q11;q12),del(9)(p?),-11,der(11)t(9;11)(p13;q12),-12,der(12)t(3;12),-13,+14,-15,-16,-17,+18,del(18)(q?)x2,der(19)t(12;19)(q13;q13)dup(12)(q24q13)t(1;12)(p22;q13),der(19)t(5;19)(q13;p13),i(19)(q10),+20,-21,der(21)t(5;21)(p?q)t(22)t(19;22)(q?q)		
Gibas et al 1984, Cancer Res		
Case No. 1	Transitional cell carcinoma	Bladder
48,XX,+i(5)(p10),+8,i(8)(q10)x2		
Case No. 6	Transitional cell carcinoma	Bladder
47,X,del(X)(q26),i(2)(p10),+i(5)(p10),del(6)(q12q21),del(10)(q24),del(13)(q12q21)		
Case No. 9	Transitional cell carcinoma	Bladder
56,Y,t(X;11)(q22;q11),+dup(1)(p11p35),add(2)(q?),+3,+4,+i(5)(p10)x2,+7,-9,-10,del(11)(p13),+12,del(13)(q12q14),der(13),+17,der(19)t(10;19)(q11;p13),-21,+der(?)t(?)t(?)p13		

- Select the number in the 'Cases' column from the results of the aberrations searcher
- Resulting page will have particular cases from publications highlighted
- Links (in orange boxes)
 - Publication information (see Slide 7-8)
 - Patient/Case information (see Slide 6)

Reference Searcher

- Search one or more reference groups for specific search criteria to retrieve relevant publications. (See slide 7)

Mitelman Reference Searcher

The Reference Database contains the complete set of references for chromosomal aberrations culled from the literature by Mitelman, Johansson, and Mertens. The references are organized into three groups:

- The Individual Cases references
- The Molecular Biology Associations references
- The Clinical Associations references

The search engine can find any reference by author, journal, year, or reference number in one, two, or all three of these groups.

Reference Searcher

Brief instructions for using the Reference Searcher:

- Check one or more of the reference group boxes.
- Enter information into one or more of the search fields.
- Click the link at Journal to view a list of all journal abbreviations.
- Press Submit Query, or press Reset to begin a new search.

Reference Group: ☐ Individual Cases
☒ Molecular Biology
☒ Clinical Association

Authors:

Journal:

Year:

Reference No.:

or

Reference List
[Full Text]
Displaying References 1 to 17 of 17 found
<p>Chng et al 2006, Leukemia</p> <p>Debes-Marun et al 2003, Leukemia</p> <p>Dewald et al 1985, Blood</p> <p>Fonseca et al 1998, Br J Haematol</p> <p>Fonseca et al 1999, Leuk Lymphoma</p> <p>Fonseca et al 2001, Blood</p> <p>Fonseca et al 2002, Blood</p> <p>Fonseca et al 2002, Blood</p> <p>Fonseca et al 2003, Blood</p> <p>Fonseca et al 2006, Leukemia</p> <p>Gertz et al 2005, Blood</p> <p>Greipp et al 2013, Br J Haematol</p> <p>Hayman et al 2001, Blood</p> <p>Hoyer et al 2000, Am J Clin Pathol</p> <p>Kapoor et al 2009, Blood</p> <p>Ludwig et al 2008, Blood</p> <p>Rajkumar et al 1999, Bone Marrow Transplant</p>

Raw Data Download

The screenshot shows the Cancer Genome Anatomy Project website. The header includes the National Cancer Institute logo and the text 'National Cancer Institute' and 'U.S. National Institutes of Health | www.cancer.gov'. The main navigation bar includes 'CGAP How To', 'Genes', 'Chromosomes', 'Tissues', 'SAGE Genie', 'RNAi', 'Pathways', and 'Tools'. The 'Chromosomes' section is active, showing a 'Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer' search interface. The 'CGAP Data' section is highlighted with a yellow box, showing a 'Download' button. The 'Purchase CGAP Reagents' section shows 'CGAP BAC Clones'. The 'Related Links' section includes 'Atlas of Genetics & Cytogenetics in Oncology & Haematology' and 'Progenetix'. The 'Mitelman Database' section provides information about the database and lists search tools: 'The Cases Quick Searcher', 'The Cases Full Searcher', 'The Molecular Biology Associations Searcher', 'The Clinical Associations Searcher', 'The Recurrent Chromosome Aberrations Searcher', and 'The Reference Searcher'. The database was last updated on February 20, 2017, with a total number of cases of 66,919 and a total number of gene fusions of 10,676.

- Mitelman Database raw data is available for download.
- Click on 'Download' under CGAP Data.
- Resulting window will have a table of information used to populate the Cancer Genome Anatomy Project site.
- Click on "mitelman.tar.gz" to download folder of raw Mitelman Database data.
- Unzip .gz file with file archiving program (i.e. 7-Zip)
- Extract all files into a folder in your desired location.
- Data can be viewed as Excel file.

Scenario #1

- You are creating a data analysis guide for a bladder cancer study your lab is conducting. You have lots of gene information, but not much on larger recurrent cytogenetic abnormalities. How can this database help?
 - Recurrent Chromosome Aberrations Searcher

Structural aberrations:



Breakpoint:

Aberration type:

All ▲

Balanced

Unbalanced ▼

Topography:

[Expand Topography List:](#)

All topographies ▲

Adrenal

Anus

Bladder

Blood vessel ▼

Morphology:

[Expand Morphology List:](#)

All morphologies ▲

Acinic cell carcinoma

Acute basophilic leukemia

Acute eosinophilic leukemia

Acute erythroleukemia (FAB type M6) ▼

Gene:

[Expand Gene List:](#)

All genes ▲

ABI1

ABL1

ACBD3

ACSL6 ▼

Retrieve

Reset Form

Numerical aberrations:



Chromosome:

Aberration type:

All ▲

Trisomy

Monosomy ▼

Cytogenetics Abnormalities in Bladder Cancer

- Tip: You may want to set a case threshold based on your results, so you're only pulling the most prevalent abnormalities for your guide.
- For example – set case threshold to 5 for rearrangements and 15 for numerical abnormalities and only pay attention to transitional cell carcinoma morphology.

Cytogenetics Abnormalities in Bladder Cancer

Balanced Chromosomal Abnormalities					
Band	Abnormality	Morphology	Topography	Cases	Genes
3p14	t(3;5)(p14;p14)	Transitional cell carcinoma	Bladder	2	
5p14	t(3;5)(p14;p14)	Transitional cell carcinoma	Bladder	2	

Unbalanced Chromosomal Abnormalities					
Band	Abnormality	Morphology	Topography	Cases	Genes
1p11	del(1)(p11)	Transitional cell carcinoma	Bladder	2	
1p22	del(1)(p22)	Transitional cell carcinoma	Bladder	4	
1q10	i(1)(q10)	Transitional cell carcinoma	Bladder	3	
1q12	del(1)(q12)	Transitional cell carcinoma	Bladder	2	
1q21	del(1)(q21)	Transitional cell carcinoma	Bladder	3	
3q21	add(3)(q21)	Transitional cell carcinoma	Bladder	2	
3q21	del(3)(q21)	Transitional cell carcinoma	Bladder	2	
3q25	del(3)(q25)	Transitional cell carcinoma	Bladder	2	
5p10	i(5)(p10)	Transitional cell carcinoma	Bladder	11	
5q11	del(5)(q11q13)	Transitional cell carcinoma	Bladder	2	
5q13	del(5)(q11q13)	Transitional cell carcinoma	Bladder	2	
5q13	del(5)(q13)	Transitional cell carcinoma	Bladder	2	
6p10	i(6)(p10)	Transitional cell carcinoma	Bladder	2	
6q13	del(6)(q13)	Transitional cell carcinoma	Bladder	2	

* Click on Number in Cases column to see evidence by publication (See slides 5-8)

Cyto Band	Abnormality	# Cases
5p10	iso(5)(p10)	11
8q10	iso(8)(q10)	8
11q10	iso(11)(q10)	5
Chr7	+7	21
Chr8	-8	17
Chr9	-9	56
Chr10	-10	16
Chr11	-11	15
Chr15	-15	17
Chr16	-16	17
Chr18	-18	17
ChrY	-Y	33

Scenario #2

- By Mate Pair sequencing, your lab has identified a $t(5;11)(p15.3;q23)$ involving NUP98 and KMT2A(MLL) in a B-cell ALL patient. Has this rearrangement been described in the literature previously?
 - Cases Quick Searcher
 - Abnormality = $t(5;11)(p15;q23)$
 - Remember: No Sub-bands!
 - Morphology = Acute lymphoblastic leukemia/lymphoblastic lymphoma

Quick Case Searcher Results

Chromosome Aberration Case List

Content-type: text/plain

[\[Full Text\]](#)

Displaying Investigations 1 to 1 of 1 found
From 1 References and 1 Cases

Swan et al 2001, Cancer Genet Cytogenet

Case No. 1 **Acute lymphoblastic leukemia/lymphoblastic lymphoma**
46,XY,t(5;11)(p15;q23)c,inv(9)(p24q32)/46,idem,add(17)(p13)

- There is one publication for this query, and it appears to be a constitutional rearrangement.



Cancer Genetics and Cytogenetics 128 (2001) 178–180

CANCER GENETICS
AND
CYTOGENETICS

Letter to the editor

Constitutional t(5;11)(p15.3;q23) in an adolescent male with acute lymphoblastic leukemia

Rearrangements involving 11q23 are well documented in hematopoietic malignancies [1]. In pediatric patients 11q23 translocations accounted for 10% of all cytogenetically abnormal acute lymphoblastic leukemia (ALL) and in 13% of ALL-L1 cases [2]. In adults, similar anomalies have been observed in 2–5% of ALL, 5–10% of de novo acute myelogenous leukemia (AML), and over 80% of topoisomerase II inhibitor-induced AML [3,4]. Almost all chromosomes in the human genome have participated as partners of 11q23 translocations [1,5–7], and in about 50–70% of cases the molecular consequence was a disruption of the mixed lineage leukemia (*MLL*) gene located in 11q23 [8–10]. However, since the function(s) of the *MLL* gene or its multiple partner genes have not been fully defined, the exact role of the resultant chimeric *MLL*-partner gene product in leukemogenesis remains uncertain. Contrary to the numerous publications on acquired 11q23 rearrangements, reports of constitutional 11q23 abnormalities in cancers are very rare. In this communication we describe a case of ALL with a constitutional 11q23 translocation.

been observed in blasts with myelomonocytic characteristics, mixed-lineage, or bi-phenotypic immunophenotype and rarely with T-cell or B-cell features [11–14]. Our patient, a carrier of a de novo constitutional t(5;11)(q15.3;q23), was diagnosed with ALL-L1 at age 16. It should be pointed out that chromosome 5 has not been documented as a translocation partner of 11q23 in hematopoietic malignancies, and we have no molecular evidence to indicate that a disruption of the *MLL* gene has taken place. Thus, whether the constitutional t(5;11)(q15.3;q23) is a coincidental feature or a predisposing factor to the development of ALL in our patient is uncertain. A search of the literature revealed two other cancer cases with constitutional chromosome 11q23 rearrangement. The first case was a one-year-old boy who presented with a unilateral retinoblastoma and a constitutional inv(11)(q23) as the only karyotypic abnormality [15]. The patient inherited the inv(11) from his father, who at the time of report was in good health. The second case involved a 25-year-old male diagnosed with B-cell diffuse lymphoma and a t(3;11)(p21;q23) as the sole karyotypic change [16].

Swan et al. 2001, PMID 11478302

Scenario #3

- How can you be sure your publications are incorporated into this database?
 - Reference Searcher
 - Search for Author: LastName FM (or F* if you don't know the middle name)

Authorship Search

Reference List

[Full Text]

Displaying References 1 to 7 of 7 found

Ahmann et al 1998, Cancer Genet Cytogenet

Dewald et al 1985, Blood

Fonseca et al 1998, Br J Haematol

Fonseca et al 2001, Blood

Hayman et al 2001, Blood

Rajkumar et al 1999, Cancer Genet Cytogenet

Woloschak et al 1986, J Cell Biochem

- Searched for “Jenkins RB”
- 7 hits in database.
- Can also search using “*” wildcard
 - Ex/ Jenkins R*

Contacts

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