

WEBLEM 3

Introduction to Nucleic Acid Databases

Introduction:

Nucleotide databases that were introduced to us in this weblem were GenBank, EMBL-EBI and DDBJ.

1. GenBank:

1. GenBank sequence database is an annotated collection of all publicly available nucleotide sequences and their protein translations.
2. This database is produced at the National Center for Biotechnology Information (NCBI) as part of an international collaboration with the European Molecular Biology Laboratory (EMBL) Data Library from the European Bioinformatics Institute (EBI) and the DNA Data Bank of Japan (DDBJ).
3. GenBank and its collaborators receive sequences produced in laboratories throughout the world from hundreds of thousands of distinct organisms.
4. GenBank continues to grow at an exponential rate, doubling every 18 months.
5. GenBank is built by direct submissions from individual laboratories and from large-scale sequencing centers.

2. EMBL-EBI:

1. It makes the world's public biological data freely available to the scientific community via a range of services and tools, perform basic research and provide professional training in bioinformatics.
2. It is part of the European Molecular Biology Laboratory (EMBL), an international, innovative and interdisciplinary research organisation funded by over 20 member states, prospect and associate member states.
3. It is situated on the Wellcome Genome Campus in Hinxton, Cambridge, UK, one of the world's large.

3. DDBJ:

1. DDBJ Center collects nucleotide sequence data as a member of INSDC(International Nucleotide Sequence Database Collaboration) and provides freely available nucleotide sequence data and supercomputer system, to support research activities in life science concentrations of scientific and technical expertise in genomics.
2. The principal purpose of DDBJ operations is to improve the quality of INSD, as public domains. When researchers make their data open to the public through INSD and commonly shared in world wide, we at DDBJ Center make efforts to describe information on the data as rich as possible, according to the unified rules of INSD, preferably without any stress by using DDBJ.

References:

1. <https://www.ncbi.nlm.nih.gov/books/NBK153518/>
2. <https://www.ebi.ac.uk/about>
3. <https://www.ddbj.nig.ac.jp/about/index-e.html>
4. <https://www.mayoclinic.org/diseases-conditions/klinefelter-syndrome/symptoms-causes/syc-20353949>

WEBLEM 3a

Introduction to Nucleic Acid Databases

Aim:

To study the genes involved in the causation of Klinefelter's syndrome using GenBank Database.

Introduction:

The query taken for Genbank is "Klinefelter's syndrome". Klinefelter syndrome is a genetic condition that results when a boy is born with an extra copy of the X chromosome.

Klinefelter syndrome is a genetic condition affecting males, and it often isn't diagnosed until adulthood. Klinefelter syndrome may adversely affect testicular growth, resulting in smaller than normal testicles, which can lead to lower production of testosterone. The syndrome may also cause reduced muscle mass, reduced body and facial hair, and enlarged breast tissue.

The effects of Klinefelter syndrome vary, and not everyone has the same signs and symptoms. About 3% of the infertile male population have Klinefelter syndrome.

Methodology:

1. Open Homepage of GenBank (URL-<https://www.ncbi.nlm.nih.gov/genbank/>)
2. Enter Search query (Klinefelter's syndrome)
3. Interpret the result and find the needed data.

Observations:

NCBI Resources How To Sign in to NCBI

GenBank Nucleotide Klinefelter Syndrome Search

GenBank Submit Genomes WGS Metagenomes TPA TSA INSDC Other

COVID-19 Information
[Public health information \(CDC\)](#) | [Research information \(NIH\)](#) | [SARS-CoV-2 data \(NCBI\)](#) | [Prevention and treatment information \(HHS\)](#) | [Español](#)

GenBank Overview

What is GenBank?

GenBank® is the NIH genetic sequence database, an annotated collection of all publicly available DNA sequences (*Nucleic Acids Research*, 2013 Jan 41(D1):D36-42). GenBank is part of the [International Nucleotide Sequence Database Collaboration](#), which comprises the DNA DataBank of Japan (DDBJ), the European Nucleotide Archive (ENA), and GenBank at NCBI. These three organizations exchange data on a daily basis.

A GenBank release occurs every two months and is available from the [ftp site](#). The [release notes](#) for the current version of GenBank provide detailed information about the release and notifications of upcoming changes to GenBank. Release notes for [previous GenBank releases](#) are also available. GenBank growth [statistics](#) for both the traditional GenBank divisions and the WGS division are available from each release.

An [annotated sample GenBank record](#) for a *Saccharomyces cerevisiae* gene demonstrates many of the features of the GenBank flat file format.

Access to GenBank

There are several ways to search and retrieve data from GenBank.

- Search GenBank for sequence identifiers and annotations with [Entrez Nucleotide](#).
- Search and align GenBank sequences to a query sequence using [BLAST](#) (Basic Local Alignment Search Tool). See [BLAST info](#) for more information about the numerous BLAST databases.
- Search, link, and download sequences programmatically using [NCBI e-utilities](#).
- The ASN.1 and flatfile formats are available at NCBI's anonymous FTP server: <ftp://ftp.ncbi.nlm.nih.gov/ncbi-asn1> and <ftp://ftp.ncbi.nlm.nih.gov/genbank>.

GenBank Data Usage

The GenBank database is designed to provide and encourage access within the scientific community to the most up-to-date and comprehensive DNA sequence information. Therefore, NCBI places no restrictions on the use or distribution of the GenBank data. However, some submitters may claim patent, copyright, or other intellectual property rights in all or a portion of the data they have submitted. NCBI is

GenBank Resources

- [GenBank Home](#)
- [Submission Types](#)
- [Submission Tools](#)
- [Search GenBank](#)
- [Update GenBank Records](#)

Fig1. Homepage of GenBank

NCBI Resources How To Sign in to NCBI

Nucleotide Nucleotide Klinefelter Syndrome Search

Create alert Advanced Help

COVID-19 Information

Public health information (CDC) | Research information (NIH) | SARS-CoV-2 data (NCBI) | Prevention and treatment information (HHS) | Español

Species Animals (4) Customize ...

Molecule types genomic DNA/RNA (6) mRNA (4) Customize ...

Source databases INSDC (GenBank) (11) RefSeq (4) Customize ...

Sequence Type Nucleotide (15)

Sequence length Custom range...

Release date Custom range...

Revision date Custom range...

Clear all Show additional filters

Summary 20 per page Sort by Default order Send to: Filters: Manage Filters

See [syndrome syndrome](#) in the Gene database syndrome reference sequences

Items: 15

☐ KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

20 bp linear DNA
Accession: DI181505.1 GI: 551423753
[Taxonomy](#)
[GenBank](#) [FASTA](#) [Graphics](#)

☐ KR 1020100093381-A/5: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

20 bp linear DNA
Accession: DI181504.1 GI: 551423752
[Taxonomy](#)
[GenBank](#) [FASTA](#) [Graphics](#)

☐ KR 1020100093381-A/4: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

20 bp linear DNA
Accession: DI181503.1 GI: 551423751
[Taxonomy](#)
[GenBank](#) [FASTA](#) [Graphics](#)

Results by taxon

Top Organisms [Tree](#)

artificial sequences (11)
synthetic construct (5)
Mus musculus (3)
Homo sapiens (1)

Analyze these sequences

Run BLAST

Find related data

Database: Select

Find items

Search details

klinefelter syndrome[All Fields]

Search See more...

Fig2. Hit page with results for query Klinefelter syndrome

NCBI Resources How To Sign in to NCBI

Nucleotide Nucleotide Advanced Help

COVID-19 Information

Public health information (CDC) | Research information (NIH) | SARS-CoV-2 data (NCBI) | Prevention and treatment information (HHS) | Español

GenBank Send to: Change region shown

Customize view

KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

GenBank: DI181505.1

[FASTA](#) [Graphics](#)

Go to: ☐

LOCUS DI181505 20 bp DNA linear PAT 23-OCT-2013

DEFINITION KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST.

ACCESSION DI181505

VERSION DI181505.1

KEYWORDS KR 1020100093381-A/6.

SOURCE artificial sequences

ORGANISM [artificial sequences](#)

other sequences.

REFERENCE 1 (bases 1 to 20)

AUTHORS Lee,S., Kim,H. and Yun,Y.

TITLE DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

JOURNAL Patent: KR 1020100093381-A 6 25-AUG-2010; College of Medicine Pochon CHA University Industry-Academic Cooperation Foundation

COMMENT KN KR 1020090012533-A/6

AN KR 1020090012533

AD 2009-02-16

PN KR 1020100093381

PD 2010-08-25

Analyze this sequence

Run BLAST

Pick Primers

Find in this Sequence

Related information

Taxonomy

Recent activity

Turn Off Clear




KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYND Nucleotide

Klinefelter Syndrome (15) Nucleotide

Klinefelter syndrome (15) Nucleotide


klinefelter syndrome (15) Nucleotide

Fig3. Result page for query Klinefelter syndrome in GenBank [KR 1020100093381-A/6]


GenBank  Send to:  Change region shown 


KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

GenBank: DI181505.1 [FASTA](#) [Graphics](#)

Go to: 

LOCUS DI181505 20 bp DNA linear PAT 23-OCT-2013
 DEFINITION KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST.
 ACCESSION DI181505
 VERSION DI181505.1
 KEYWORDS KR 1020100093381-A/6.
 SOURCE artificial sequences
 ORGANISM [artificial sequences](#)
 other sequences.
 REFERENCE 1 (bases 1 to 20)
 AUTHORS Lee,S., Kim,H. and Yun,Y.
 TITLE DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST
 JOURNAL Patent: KR 1020100093381-A 6 25-AUG-2010;
 College of Medicine Pochon CHA University Industry-Academic Cooperation Foundation
 COMMENT KN KR 1020090012533-A/6
 AN KR 1020090012533
 AD 2009-02-16
 PN KR 1020100093381
 PD 2010-08-25

Analyze this sequence 
 Run BLAST
 Pick Primers
 Find in this Sequence

Related information 
 Taxonomy



Recent activity 
 Turn Off Clear
 KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYND Nucleotide
 Klinefelter Syndrome (15) Nucleotide
 Klinefelter syndrome (15) Nucleotide
 klinefelter syndrome (15) Nucleotide

Fig4. Header of result for query Klinefelter syndrome [KR 1020100093381-A/6]

[FASTA](#) [Graphics](#)


Go to: 


LOCUS DI181505 20 bp DNA linear PAT 23-OCT-2013
 DEFINITION KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST.
 ACCESSION DI181505
 VERSION DI181505.1
 KEYWORDS KR 1020100093381-A/6.
 SOURCE artificial sequences
 ORGANISM [artificial sequences](#)
 other sequences.
 REFERENCE 1 (bases 1 to 20)
 AUTHORS Lee,S., Kim,H. and Yun,Y.
 TITLE DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST
 JOURNAL Patent: KR 1020100093381-A 6 25-AUG-2010;
 College of Medicine Pochon CHA University Industry-Academic Cooperation Foundation
 COMMENT KN KR 1020090012533-A/6
 AN KR 1020090012533
 AD 2009-02-16
 PN KR 1020100093381
 PD 2010-08-25
 AT DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST AI Lee, Su-Man|Kim, Hwan-Hee|Yun, Yeo-Jin
 AA College of Medicine Pochon CHA University Industry-Academic AA Cooperation Foundation
 PR
 OS artificial sequences
 TY DNA.

FEATURES
 source Location/Qualifiers
 1..20
 /organism="artificial sequences"
 /mol_type="unassigned DNA"
 /db_xref="taxon:81077"

ORIGIN
 1 gcgggtcacac aggaaaagat
 //

Run BLAST
 Pick Primers
 Find in this Sequence

Related information 
 Taxonomy

Recent activity 
 Turn Off Clear
 KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYND Nucleotide
 Klinefelter Syndrome (15) Nucleotide
 Klinefelter syndrome (15) Nucleotide
 klinefelter syndrome (15) Nucleotide
 Klinefelters syndrome (0) Nucleotide
 See more...

You are here: NCBI > DNA & RNA > Nucleotide Database Support Center

Fig5. Result page of query Klinefelter syndrome in GenBank with Features and Origin shown

FASTA ▾

Send to: ▾

Change region shown ▾

Customize view ▾

KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST

GenBank: DI181505.1

[GenBank](#) [Graphics](#)

>DI181505.1 KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST
GCGGTCACACAGGAAAAGAT

Analyze this sequence ▴[Run BLAST](#)[Pick Primers](#)[Find in this Sequence](#)**Related information** ▴[Taxonomy](#)**Recent activity** ▴[Turn Off](#) [Clear](#)

KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYND Nucleotide

Klinefelter Syndrome (15) Nucleotide

Klinefelter syndrome (15) Nucleotide

klinefelter syndrome (15) Nucleotide

Klinefelters syndrome (0) Nucleotide

[See more...](#)

Fig6. FASTA sequence of Result of query Klinefelter syndrome in GenBank [KR 1020100093381-A/6]

NCBI Resources How To Sign in to NCBI

Nucleotide Nucleotide ▾ Klinefelter Syndrome Create alert Advanced Search Help

COVID-19 Information

[Public health information \(CDC\)](#) | [Research information \(NIH\)](#) | [SARS-CoV-2 data \(NCBI\)](#) | [Prevention and treatment information \(HHS\)](#) | [Español](#)

Species Customize ...

Molecule types genomic DNA/RNA (6) Customize ...

Source databases clear

- ☒ **INSDC (GenBank) (11)**
- ☐ RefSeq (0)
- ☐ Customize ...

Sequence Type Nucleotide (11)

Sequence length Custom range...

Release date Custom range...

Revision date Custom range...

[Clear all](#)

[Show additional filters](#)

Summary ▾ 20 per page ▾ Sort by Default order ▾

See [syndrome syndrome](#) in the Gene database syndrome reference sequences

Items: 11

Filters activated: INSDC (GenBank). [Clear all](#)

- ☐ [KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST](#)
20 bp linear DNA
Accession: DI181505.1 GI: 551423753
[Taxonomy](#)
[GenBank](#) [FASTA](#) [Graphics](#)
- ☐ [KR 1020100093381-A/5: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST](#)
20 bp linear DNA
Accession: DI181504.1 GI: 551423752
[Taxonomy](#)
[GenBank](#) [FASTA](#) [Graphics](#)
- ☐ [KR 1020100093381-A/4: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST](#)
20 bp linear DNA
Accession: DI181503.1 GI: 551423751
[Taxonomy](#)

Filters: [Manage Filters](#)

Results by taxon ▴

Top Organisms [\[Tree\]](#)

- artificial sequences (11)
- synthetic construct (5)

Analyze these sequences ▴

[Run BLAST](#)

Find related data ▴

Database: Select ▾

[Find items](#)

Search details ▴

klinefelter syndrome[All Fields]
AND ddbj_embl_genbank[filter]

[Search](#) [See more...](#)

Fig7. Hit page for Klinefelter syndrome in GenBank with limit options and refined results

NCBI
Resources
How To
Sign in to NCBI

Nucleotide Home
Help

Nucleotide Advanced Search Builder

i Filters activated: INSDC (GenBank). [Clear all](#)

Use the builder below to create your search

[Edit](#)
[Clear](#)

Builder

All Fields

AND
All Fields

Search
or [Add to history](#)

History
[Download history](#)
[Clear history](#)

Search	Add to builder	Query	Items found	Time
#8	Add	Search Klinefelter Syndrome Filters: INSDC (GenBank)	11	15:37:38
#6	Add	Search Klinefelter Syndrome	15	15:16:23
#5	Add	Search Klinefelter syndrome	15	14:53:39
#2	Add	Search klinefelter syndrome	15	14:53:25
#4	Add	Search Klinefelters syndrome	0	14:53:12
#3	Add	Search Klinefelter's syndrome	2	14:52:49

Fig8. Advanced search builder for Query Klinefelter Syndrome in GenBank

Conclusion:

GenBank is an amazing tool integrated within NCBI to make the querying of nucleotide sequences easy and accesible. The dataset is vast and depending on the query thousands of results can be poured over in a very short amount of time.

According, to the current time GenBank also provide detailed information about the release and notifications of upcoming changes in GenBank.

Result:

Sr. No.	Resource	Result
1	GenBank	15

References:

- <https://www.ncbi.nlm.nih.gov/genbank/>
- <https://www.ncbi.nlm.nih.gov/nuccore/?term=Klinefelter+Syndrome>
- <https://www.ncbi.nlm.nih.gov/nuccore/DI181505.1>
- <https://www.ncbi.nlm.nih.gov/nuccore/DI181505.1?report=fasta>

WEBLEM 3b

Introduction to Nucleic Acid Sequence Database

Aim:

To study the genes involved in the causation of Klinefelter's syndrome using EMBL-EBI Database.

Introduction:

The query taken for EMBL-EBI is "Klinefelter's syndrome". Klinefelter syndrome is a genetic condition that results when a boy is born with an extra copy of the X chromosome.

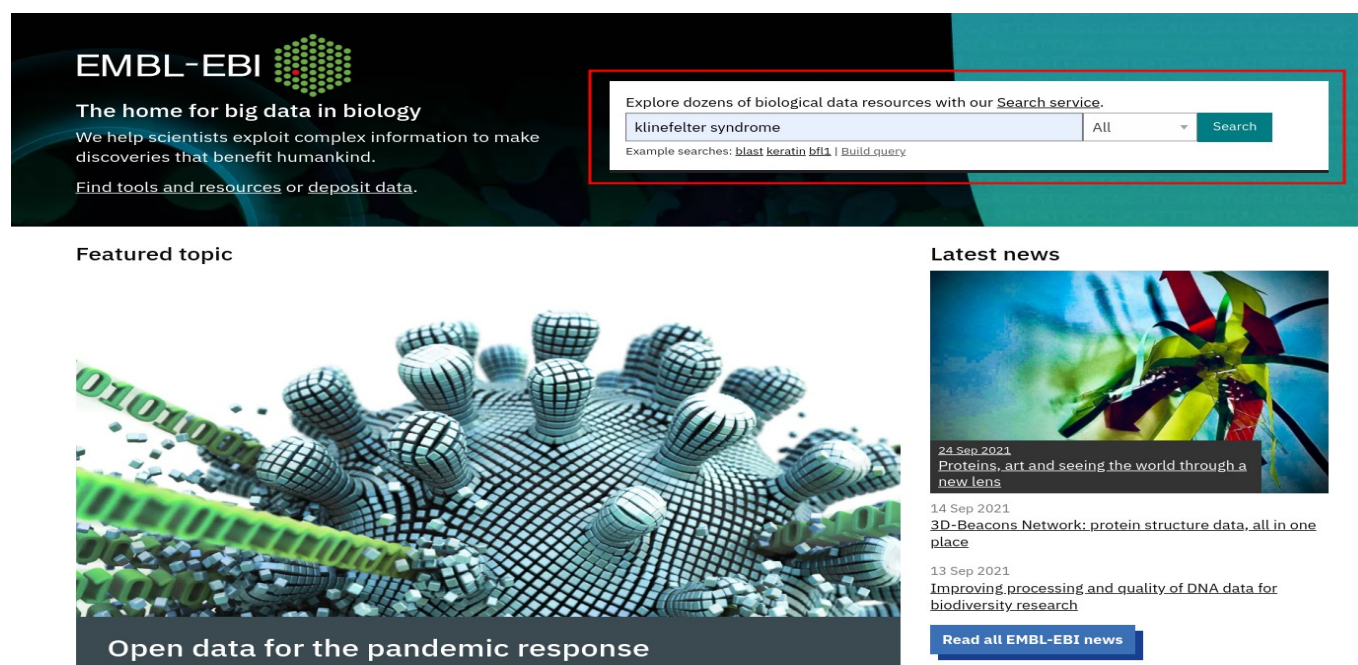
Klinefelter syndrome is a genetic condition affecting males, and it often isn't diagnosed until adulthood. Klinefelter syndrome may adversely affect testicular growth, resulting in smaller than normal testicles, which can lead to lower production of testosterone. The syndrome may also cause reduced muscle mass, reduced body and facial hair, and enlarged breast tissue.

The effects of Klinefelter syndrome vary, and not everyone has the same signs and symptoms. About 3% of the infertile male population have Klinefelter syndrome.

Methodology:

1. Open Homepage of EMBL-EBI (URL-<https://www.ebi.ac.uk/>)
2. Enter Search query (Klinefelter's syndrome)
3. Interpret the result and find the needed data.

Observation:



The screenshot shows the EMBL-EBI homepage. At the top left, the EMBL-EBI logo is displayed with the tagline "The home for big data in biology". Below this, a search bar is highlighted with a red box, containing the text "klinefelter syndrome". To the right of the search bar is a "Search" button. Below the search bar, there are example searches: "blast", "keratin", "bfl1", and a link to "Build query".

Below the search bar, the page is divided into two main sections: "Featured topic" and "Latest news".

Featured topic: This section features a large, stylized image of a DNA double helix and a protein structure. Below the image, the text "Open data for the pandemic response" is displayed.

Latest news: This section lists three news items:

- 24 Sep 2021: Proteins, art and seeing the world through a new lens
- 14 Sep 2021: 3D-Beacons Network: protein structure data, all in one place
- 13 Sep 2021: Improving processing and quality of DNA data for biodiversity research

At the bottom of the "Latest news" section, there is a blue button labeled "Read all EMBL-EBI news".

Fig1. Homepage of EMBL with query Klinefelter entered in the search bar

Search results for *kliefelter syndrome*

Showing 18 results out of 6,196 in All results

Filter your results

Source

All results (6,196)

- Genomes & metagenomes (1)
- Nucleotide sequences (178)
- Protein sequences (1)
- Gene expression (3,832)
- Diseases (33)
- Protein expression data (1)
- Literature (1,978)
- Samples & ontologies (172)

Gene expression (3,832 results)

Source: ArrayExpress (ID: E-GEOD-37258)

Expression data of the iPSCs derived from foreskin fibroblast cells of normal person and KS patient

Klinefelter's **syndrome** (KS) is one of the common chromosome aneuploidy diseases in males with unexplained physiological mechanism. iPSCs, are similar to ESCs in terms of indefinite self-renewal and pluripotency, provided an alternative choice for modeling disease to facilitate the disease ...

Cross References: Gene expression (1) Literature (1) Samples & ontologies (1)

Source: ArrayExpress (ID: E-MTAB-4922)

Transcription profiling by array of eight human **Klinefelter syndrome** patients (47,XXY) and eight human controls

Klinefelter syndrome (KS) is the most common male chromosomal abnormality (47,XXY). Males with KS suffers from numerous comorbidities. Gene expression data integrated with other data types can ... **Klinefelter syndrome** patients (47,XXY) and eight human controls. The age of the **Klinefelter syndrome**

Fig2. Hit page of EMBL for query Klinefelter Syndrome

Search results for *kliefelter syndrome*

Showing 15 results out of 178 in All results → Nucleotide sequences

Filter your results

Save result

Create RSS feed

Source

All results (6,196)

Nucleotide sequences (178)

- ENA Study (17)
- Sequence (11)
- Study (Read/Analysis) (11)
- Sample (23)
- Read (Run) (58)
- Read (Experiment) (58)

☐ Nucleotide sequences (178 results)

Source: ENA Study (ID: PRJEB22968)

☐ Decreased miRNA expression in **Klinefelter syndrome**

Decreased miRNA expression in **Klinefelter syndrome**

Cross References: Nucleotide sequences (41) Samples & ontologies (1)

Organisms

- ☐ Homo sapiens (153)
- ☐ artificial sequences (2)
- ☐ synthetic construct (5)
- ☐ Mus musculus (3)

Source: Sample (ID: SRS2498281)

☐ SAMN07615671

fetal **Klinefelter syndrome** 7

Cross References: Nucleotide sequences (3) Samples & ontologies (2)

Fig3. Hit page of nucleotide sequences for Klinefelter syndrome

EMBL-EBI Services Research Training About us EMBL-EBI

ENA
European Nucleotide Archive

Enter text search terms Search
Examples: histone, BN000065
PRJEB22968 View
Examples: Taxon:9606, BN000065, PRJEB402

Home Submit Search Rulespace About Support

Project: PRJEB22968

The widely variable phenotypic spectrum and the different symptom severity in men with Klinefelter syndrome (KS) suggest a role for epigenetic mediators. Therefore, the aim of this study was to evaluate the possible involvement of miRNAs in the clinical manifestations of KS. To accomplish this, we performed a transcriptome analysis in peripheral blood mononuclear cells (PBMCs) of 10 non-mosaic KS patients, 10 aged-matched healthy male and 10 aged-matched healthy female controls with normal karyotype. After RNA extraction from PBMC and the preparation of small RNA libraries, the samples were sequenced using next generation high-throughput sequencing technology. Expression profiling analysis revealed a significant differential expression of 2 miRNAs in KS compared to male controls. In particular, MIR3648 resulted significantly ($p < 0.0001$) down-regulated by -19.084-fold, while MIR3687 was non-expressed at all ($p < 0.0001$) in KS patients. These results were confirmed by qRT-PCR. The functional analysis of the two transcripts showed that they seem to play a role in breast cancer, hemopoietic abnormalities, immune defects and adipocyte differentiation and fat cell maturation. Therefore, we speculate that both miRNAs may play a role in the immune and metabolic disorders and in the risk of breast cancer development in men with KS.

Show More

View: XML
XML (STUDY)

Download: XML
XML (STUDY)

Navigation: Show

Read Files: Hide

Related ENA Records: Show

Secondary Study Accession: ERP104698

Study Title: Decreased miRNA expression in Klinefelter syndrome

Center Name: Genomix4Life

Study Name: Decreased miRNA expression in Klinefelter syndrome

Broker Name: ArrayExpress

ENA-FIRST-PUBLIC: 2017-12-02

ENA-LAST-UPDATE: 2017-10-10

Show More

Read Files

Fig4. Result page of nucleotide sequences for Klinefelter's syndrome [Project: PRJEB22968]

EMBL-EBI Services Research Training About us EMBL-EBI

EBI Search

klinefelter syndrome
Examples: VAV_HUMAN, tp53, Sulston... Build Query

Help & Documentation About EBI Search ORCID data claiming Feedback

Search results for *klinefelter syndrome*

Showing 15 results out of 3,832 in All results → Gene expression

Filter your results

Source
All results (6,196)
Gene expression (3,832)
ArrayExpress (10)
Expression Atlas Experiments (1)
Differential Expression Atlas Genes (3,814)
dbGaP (1)
GEO (6)

Organisms
☐ Homo sapiens (3,824)
☐ Mus musculus (3)

☐ **Gene expression** (3,832 results)

Source: ArrayExpress (ID: E-GEOD-37258)

☐ Expression data of the iPSCs derived from foreskin fibroblast cells of normal person and KS patient

Klinefelter's **Syndrome** (KS) is one of the common chromosome aneuploidy diseases in males with unexplained physiological mechanism. iPSCs, are similar to ESCs in terms of indefinite self-renewal and pluripotency, provided an alternative choice for modeling disease to facilitate the disease ...

Cross References: Gene expression (1) Literature (1) Samples & ontologies (1)

Source: ArrayExpress (ID: E-MTAB-4922)

☐ Transcription profiling by array of eight human **Klinefelter syndrome** patients (47,XXY) and eight human controls

Klinefelter syndrome (KS) is the most common male chromosomal abnormality (47,XXY). Males with KS suffers from numerous comorbidities. Gene

Fig5. Hit page of Gene expression for Klinefelter Syndrome

ARRAYEXPRESS / BROWSE / E-GEOD-37258

Please note that we have stopped the regular imports of Gene Expression Omnibus (GEO) data into ArrayExpress. This may not be the latest version of this experiment.

E-GEOD-37258 - Expression data of the iPSCs derived from foreskin fibroblast cells of normal person and KS patient

Status	Released on 13 April 2012, last updated on 28 September 2015
Organism	Homo sapiens
Samples (18)	Click for detailed sample information and links to data
Array (1)	A-AFFY-44 - Affymetrix GeneChip Human Genome U133 Plus 2.0 [HG-U133_Plus_2]
Protocols (7)	Click for detailed protocol information
Description	Klinefelter's Syndrome (KS) is one of the common chromosome aneuploidy diseases in males with unexplained physiological mechanism. iPSCs, are similar to ESCs in terms of indefinite self-renewal and pluripotency, provided an alternative choice for modeling disease to facilitate the disease research in vitro. We used microarray to detect the global reprogramming of KS and normal fibroblast cells to iPSCs. Also we used microarray to explore the possible molecular varieties between KS patient and normal person in the early development. Fibroblast cells from both normal person and KS patient were reprogrammed into iPSCs by ectopic expression of OCT4, SOX2, KLF4 and C-MYC. The expression profiles of normal and KS fibroblast cells, a line of normal iPSCs and two lines of KS iPSCs as well as a line of human ESCs were detected.
Experiment type	transcription profiling by array
Contacts	Yu Ma <nkmy13@163.com>, Ying Jin
Citation	Aberrant gene expression profiles in pluripotent stem cells induced from fibroblasts of a Klinefelter syndrome patient . Ma Y, Li C, Gu J, Tang F, Li C, Li P, Ping P, Yang S, Li Z, Jin Y.
MIAME	

Fig6. Result page of Gene Expression for Klinefelter Syndrome [E-GEOD-37258]

Search results for *kliefelter syndrome*

Showing 15 results out of 33 in All results → Diseases

Filter your results

Source

All results (6,196)

Diseases (33)

OMIM (31)

VarSite (2)

Save result

Create RSS feed

☐ Diseases (33 results)

Source: OMIM (ID: 300228)

☐ TESTICULAR GERM CELL TUMOR 1

... familial male breast cancer. MOLECULAR GENETICS - TGCT Locus and **Klinefelter Syndrome** Rapley et al. (2000) pointed out that **Klinefelter syndrome** (47,XXY) is a risk factor for extragonadal germ cell tumors. The relative risk of mediastinal germ cell tumors in **Klinefelter syndrome** is 67 (Has ...

Cross References: Diseases (3)

Source: OMIM (ID: 254000)

☐ MUSCULAR DYSTROPHY, CONGENITAL, WITH INFANTILE CATARACT AND HYPOGONADISM

Bassoe (1956) described a **syndrome** of congenital muscular dystrophy, infantile cataract, and hypogonadism (in females ovarian agenesis, in males **Klinefelter syndrome**). Seven persons living in a small, isolated Norwegian village were identified.

Fig7. Hit page of Diseases for Klinefelter's syndrome

Search OMIM...

Q

Options

%300228

Table of Contents

Title

Gene-Phenotype Relationships

Text

Mapping

Molecular Genetics

References

Contributors

Creation Date

Edit History

% 300228

TESTICULAR GERM CELL TUMOR 1; TGCT1

Cytogenetic location: *Xq27* Genomic coordinates (GRCh38): *X:143,000,000-148,000,000*

Gene-Phenotype Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
<i>Xq27</i>	Testicular germ cell tumor	300228		2

TEXT

For a general phenotypic description and a discussion of genetic heterogeneity of testicular germ cell tumors, see 273300.

Mapping

Rapley et al. (2000) analyzed the X chromosome in 99 families with testicular germ cell tumor compatible with X linkage, 80% of which were sib pairs, and found preliminary evidence for a TGCT predisposition locus at Xq27-q28 (hloc score, 2.01). Stratification analysis showed that families with at least 1 case of bilateral disease showed strong evidence of linkage to the Xq27 locus, with an hloc score of 4.76 (genomewide p = 0.034). The proportion of families with undescended testis linked to this locus was 73% compared with 26% of families without undescended testis. These results provided evidence for a TGCT susceptibility gene on Xq27 that may also predispose to undescended testis. Rapley et al. (2000) stated that TGCT1 was the first cancer susceptibility gene to be mapped in a genomewide search predominantly using sib pairs and the third cancer-predisposing gene to be mapped to the X chromosome, the others being the prostate cancer susceptibility gene (HPCX; 300147) and the androgen receptor gene (AR; 313700), mutations in which are associated with familial male breast cancer.

External Links

Gene Info

Clinical Resources

Clinical Trials

Animal Models

Fig8. Result page of Diseases for Klinefelter Syndrome [TESTICULAR GERM CELL TUMOR 1; TGCT1]

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EBI Search

klinefelter syndrome

Examples: VAV_HUMAN, tp53, Sulston...

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Search results for **klinefelter syndrome**

Showing 15 results out of 1,978 in All results → Literature

Filter your results

Source

All results (6,196)

Literature (1,978)

Europe PMC (1,978)

Publication Date

☐ 2021 (99)
☐ 2020 (142)
☐ 2019 (106)
☐ 2018 (108)
☐ 2017 (86)
☐ 2016 (77)
☐ 2015 (91)
☐ 2014 (92)
☐ 2013 (79)

Save result

Create RSS feed

☐ Literature (1,978 results)

Sort by Relevance

Source: Europe PMC (ID: 24910838)

☐ Unusual presentation of **Klinefelter syndrome**.
Das C, Sahana PK, Sengupta N, Roy M, Dasgupta R
(2013 Dec 01), *Indian journal of endocrinology and metabolism* 17 (Suppl 3) S683-4

Source: Europe PMC (ID: 24786702)

☐ **Klinefelter syndrome** and TESE-ICSI.
Plotton I, Brosse A, Cuzin B, Lejeune H
(2014 Apr 29), *Annales d'endocrinologie* 75 (2) 118-125

Cross References: Samples & ontologies (19)

Fig9. Hit page of literature for Klinefelter's Syndrome

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Europe PMC

Search life-sciences literature (39,361,030 articles, preprints and more)

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Advanced search

Quick link: Coronavirus articles and preprints

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Abstract
Figures (2)
Free full text

Similar Articles

Unusual presentation of Klinefelter syndrome.

Das C¹, Sahana PK¹, Sengupta N¹, Roy M¹, Dasgupta R¹

Author information

Indian Journal of Endocrinology and Metabolism, 01 Dec 2013, 17(Suppl 3):S683-4
DOI: 10.4103/2230-8210.123567 PMID: 24910838 PMCID: PMC4046601

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Share this article

Annotations (50)
Get citation
Claim to ORCID

Abstract

Introduction

Klinefelter syndrome usually presents in the puberty and adulthood with its characteristic features. We report a boy who had Klinefelter syndrome with hypospadias and hydrocele.

Case note

Six and half year old boy had complaints of genitourinary problem in the form of hypospadias, small phallus and hydrocele. Karyotyping showed 47,XXY.

Conclusion

This case illustrates that Klinefelter syndrome was presented in the infancy with hypospadias and hydrocele which

Fig10. Result page of Literature for Klinefelter’s syndrome [Unusual presentation of Klinefelter syndrome]

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EBI Search

Build Query

Examples: VAV_HUMAN, tp53, Sulston...

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[ORCID data claiming](#)

[Feedback](#)

Search results for **klinefelter syndrome**

Showing 15 results out of 172 in All results → Samples & ontologies

Filter your results

Source
All results (6,196)
Samples & ontologies (172)
BioSamples (43)
EFO (2)
MESH (1)
Ontology Lookup Service (OLS) (13)
BioStudies (113)

☐ Samples & ontologies (172 results)

Source: BioStudies (ID: S-EPMC5469740)

☐ Is there any clinical relevant difference between non mosaic **Klinefelter Syndrome** patients with or without Androgen Receptor variations?

Klinefelter Syndrome (KS) is the most common chromosomal disorder in men leading to non-obstructive azoospermia. Spermatozoa can be found by TESE in about 50% of adults with KS despite severe ... variation and a synonymous variation not leading to amino acid substitution. All the **Klinefelter** ...

Source: BioStudies (ID: S-EPMC5413098)

☐ Effects of Oxandrolone on Cardiometabolic Health in Boys With **Klinefelter Syndrome**: A Randomized Controlled Trial.

Context: **Klinefelter syndrome** (KS) is a common condition in males, resulting in androgen deficiency and cardiometabolic diseases. These

Fig11. Hit page of Samples and ontologies for klinefelter’s syndrome

BIOSTUDIES / STUDIES / S-EPMC5469740

Release Date: 1 January 2017

{JSON} <XML> PageTab FTP

Is there any clinical relevant difference between non mosaic Klinefelter Syndrome patients with or without Androgen Receptor variations?

Valente U¹, Vinanzi C¹, Dipresa S¹, Selice R¹, Menegazzo M¹, Iafrate M¹, Foresta C¹, Garolla A²

¹ Unit of Andrology and Reproductive Medicine, Department of Medicine, University of Padova, 35122, Padova, PD, Italy. ² Unit of Andrology and Reproductive Medicine, Department of Medicine, University of Padova, 35122, Padova, PD, Italy. andrea.garolla@unipd.it.

Accession S-EPMC5469740

Abstract Klinefelter Syndrome (KS) is the most common chromosomal disorder in men leading to non-obstructive azoospermia. Spermatozoa can be found by TESE in about 50% of adults with KS despite severe testicular degeneration. We evaluated AR variations and polymorphism length in 135 non-mosaic KS patients, aimed to find possible correlation with clinical features, sex hormones and sperm retrieval. Among 135 KS patients we found AR variations in eight subjects (5.9%). All variations but one caused a single amino acid substitution. Four variations P392S, Q58L, L548F, A475V found in six patients had been previously described to be associated with different degrees of androgen insensitivity. Moreover we observed in two patients Y359F and D732D novel variations representing respectively a missense variation and a synonymous variation not leading to amino acid substitution. All the Klinefelter patients with AR gene variations were azoospermic. Spermatozoa were retrieved with TESE for two men (40%), sperm retrieval was unsuccessful in other 3 patients. This is the only study reporting AR variations in KS patients. Relevant clinical differences not emerged between AR mutated and not AR mutated KS patients, but does each variation play an important role in the transmission to the offspring obtained by ART in this patients?

Publication *Scientific reports*. 2017 Jun; Volume 7(1). Page: 3358. doi: 10.1038/s41598-017-03371-y

PMCID: PMC5469740 PMID: 28611373

Linked Information

Show 5 entries Search:

Name	Type
NM_000044.2	Nucleotide

Showing 1 to 1 of 1 entries

ORCID: Data claiming

You can sign-in to ORCID to claim your data

☐ Remember me on this computer

Similar Studies

- High-resolution analysis of germ cells from men with sex chromosomal aneuploidies reveals normal transcriptome but impaired imprinting. [S-EPMC5724305]
- Testis Transcriptome Modulation in Klinefelter Patients with Hypospermatogenesis. [S-EPMC5374630]
- The risk of TESE-induced hypogonadism: a

Fig12. Result page of Samples and entologies for Klinefelter's Syndrome [S-EPMC5469740]

Conclusion:

EMBL provides a big combination of databases to choose from just like NCBI does. We can use it to found almos all kinds of relevant information related to research topic

Results:

Sr. No	Resource	Results
1	Nucleotide Sequences	178
2	Gene Expression	3832
3	Diseases	33
4	Literature	1978
5	Samples and entologies	172

References:

- https://www.ebi.ac.uk/ebisearch/search.ebi?query=klinefelter%20syndrome&db=allebi&requestFrom=ebi_index
- Nucleotide Sequence**
 - <https://www.ebi.ac.uk/ebisearch/search.ebi?db=nucleotideSequences&query=klinefelter%20syndrome>

2. <https://www.ebi.ac.uk/ena/browser/view/PRJEB22968?show=reads>

3. Gene Expression

1. <https://www.ebi.ac.uk/arrayexpress/experiments/E-GEOD-37258>

4. Diseases

1. <https://www.ebi.ac.uk/ebisearch/search.ebi?db=diseases&query=klinefelter%20syndrome>

2. <https://www.omim.org/entry/300228>

5. Literature

1. <https://www.ebi.ac.uk/ebisearch/search.ebi?db=literature&query=klinefelter%20syndrome>

2. <https://europepmc.org/article/MED/24910838>

6. Samples and ontologies

1. <https://www.ebi.ac.uk/ebisearch/search.ebi?db=ontologies&query=klinefelter%20syndrome>

2. <https://www.ebi.ac.uk/biostudies/studies/S-EPMC5469740>

WEBLEM 3c

Introduction to Nuclei Acid Sequence Database

Aim:

To study the genes involved in the causation of Klinefelter's syndrome using DDBJ Database..

Introduction:

The query taken for DDBJ is "Klinefelter's syndrome". Klinefelter syndrome is a genetic condition that results when a boy is born with an extra copy of the X chromosome.

Klinefelter syndrome is a genetic condition affecting males, and it often isn't diagnosed until adulthood. Klinefelter syndrome may adversely affect testicular growth, resulting in smaller than normal testicles, which can lead to lower production of testosterone. The syndrome may also cause reduced muscle mass, reduced body and facial hair, and enlarged breast tissue.


The effects of Klinefelter syndrome vary, and not everyone has the same signs and symptoms. About 3% of the infertile male population have Klinefelter syndrome.

Methodology:


4. Open Homepage of DDBJ (URL-<https://www.ddbj.nig.ac.jp/index-e.html>)
5. Navigate to Services > Getentry
6. Enter Search query (Accession ID of a paper based on Klinefelter Syndrome)
7. Interpret the result and find the needed data.

Observations:

DDBJ/BioProject/BioSample/DRA/GEA submission services disruption




Bioinformation and DDBJ Center provides sharing and analysis services for data from life science researches and advances science.




Services

Search, analysis, database services of DDBJ Center




Super Computer

NIG Supercomputer




Statistics

Statistics of DDBJ Center services



Activities

Training sessions and achievements of DDBJ Center



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NEWS

DDBJ/BioProject/BioSample/DRA/GEA submission services disruption

2021/09/15 [Announcement](#) [DDBJ](#) [BioProject](#) [BioSample](#) [DRA](#) [GEA](#) [DDBJ Center](#)

The Report for the 34th International Collaborators Meeting

2021/09/10 [Announcement](#) [DDBJ](#) [DDBJ Center](#)

Release of SARS-CoV-2 sequence data (*Severe acute respiratory syndrome coronavirus 2*)

2021/09/09 [Data Release](#) [DDBJ](#) [DDBJ Center](#)

Sequence data release of a species of chrysanthemum (cultivar name Ginyose) (*Chrysanthemum seticuspe*)

2021/09/03 [Data Release](#) [DDBJ](#) [BioProject](#) [BioSample](#) [DRA](#) [DDBJ Center](#)

Sequence data release of mangrove cricket (*Apteronomobius asahinai*)

2021/09/03 [Data Release](#) [DDBJ](#) [BioProject](#) [BioSample](#) [DRA](#) [DDBJ Center](#)


more 

Fig1. Homepage of DDBJ

analysis 7
submission 10
annotation 1
DDBJ 21
DBCLS 8
BINDS 1
NBDC 2

JGA
database submission DDBJ
JGA is a controlled-access database for sharing individual-level genotype and phenotype information.

MSS
submission DDBJ
Submission of large-scale data such as WGS, complete genome and TSA.

NSSS
submission DDBJ
Interactive nucleotide sequence submission system via web wizard

TogoAnnotator
search DBCLS
This tool normalizes gene product names and assists with the curation task.

WABI
search analysis DDBJ
WABI is a web API for using the search services of the DDBJ without needing to navigate the web front-end

JVar
database submission DDBJ
JVar archives human genome variations.

Maser
analysis BINDS
Maser is a web based analysis environment especially for data from Next Generation Sequencers. This service has been developed by Cell Innovation Program etc.

RefEx
search DBCLS
A web tool for a comfortable search of reference data for gene expression analysis

TogoVar
search NBDC
A database collects and organizes genome sequence differences between individuals (variants) in the Japanese population and disease information associated with.

getentry
search DDBJ
DDBJ annotated/assembled data retrieval by accession numbers

MAFFT
analysis DDBJ
Fast multiple alignment tool for nucleotide and amino acid sequences.

NHA
database NBDC
A platform for sharing various data generated from human specimens

TXSearch
search DDBJ
Taxonomy database search

VecScreen
analysis DDBJ
Vector search to screen contamination in nucleic acid sequences.

Fig2. Services page of DDBJ with getentry highlighted



About DDBJ
How to Use
Report/Statistics
FAQ

HOME > Search and Analysis > getentry

getentry

Data retrieval by accession numbers etc

ID :

DNA Database : ☒ DDBJ / EMBL / GenBank ☐ MGA Output Format :

Protein Database : ☐ UniProt ☐ PDB ☐ DAD ☐ Patent Output Format :

Result : Limit : Results

Fig3. Getentry page of DDBJ with accession ID of result from GenBank [Weblem 3a] entered

LOCUS	DI181505	20 bp	DNA	linear	PAT 23-OCT-2013
DEFINITION	KR 1020100093381-A/6: DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST.				

HEADER

ACCESSION [DI181505](#)
 VERSION DI181505.1
 KEYWORDS KR 1020100093381-A/6.
 SOURCE artificial sequences
 ORGANISM [artificial sequences](#)
 other sequences.
 REFERENCE 1 (bases 1 to 20)
 AUTHORS Lee,S., Kim,H. and Yun,Y.
 TITLE DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST
 JOURNAL Patent: KR 1020100093381-A 6 25-AUG-2010;
 College of Medicine Pochon CHA University Industry-Academic Cooperation Foundation
 COMMENT KN KR 1020090012533-A/6
 AN KR 1020090012533
 AD 2009-02-16
 PN KR 1020100093381
 PD 2010-08-25
 AT DIAGNOSTIC METHOD OF KLINEFELTER SYNDROME BY QUANTITATING THE PROMOTER AND 5'-END METHYLATION STATUS OF XIST
 AI Lee, Su-Man|Kim, Hwan-Hee|Yun, Yeo-Jin
 AA College of Medicine Pochon CHA University Industry-Academic Cooperation Foundation
 PR
 OS artificial sequences
 TY DNA

FEATURES

FEATURES	Location/Qualifiers
source	1..20 /mol_type="unassigned DNA" /db_xref="taxon:81077" /organism=" artificial sequences "

BASE COUNT	8 a	4 c	6 g	2 t
------------	-----	-----	-----	-----

ORIGIN	1 gcggtcacac aggaaaagat
//	

ORIGIN

Fig4. Result page with Header, Features and Origin Highlighted for Klinefelter Syndrome

Conclusion:

DDBJ provides complete data on a nucleotide sequence based on the accession ID of the sequence. It is an easy and quick way to cross check and identify the necessary data

Result:

On firing the accession ID for the Klinefelter Syndrome sequence was retrieved

References:

1. <https://www.ddbj.nig.ac.jp/index-e.html>
2. <https://www.ddbj.nig.ac.jp/services/index-e.html>
3. <http://getentry.ddbj.nig.ac.jp/top-e.html>
4. http://getentry.ddbj.nig.ac.jp/getentry/na/DI181505/?format=flatfile&filetype=html&trace=true&show_suppressed=false&limit=10