Introduction to the UCSC genome browser

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What we will cover

Structure of the human genome

Genomic information

Data acquisition

UCSC Genome Browser
Structure of human genome

1. At the simplest level, chromatin is a double-stranded helical structure of DNA.
2. DNA is complexed with histones to form nucleosomes.
3. Each nucleosome consists of eight histone proteins around which the DNA wraps 1.65 times.
4. A chromatome consists of a nucleosome plus the H1 histone.
5. The nucleosomes fold up to produce a 30-nm fiber...
6. ...that forms loops averaging 300 nm in length.
7. The 300-nm fibers are compressed and folded to produce a 250-nm-wide fiber.
8. Tight coiling of the 250-nm fiber produces the chromatid of a chromosome.

Structure of human genome

• Total of 23 pairs of chromosomes.
• Each chromosome is diploid.
• Each individual chromosome made up of double stranded DNA.
• ~3 billion bps (2m) compacted in a cell (15 μm)
Information in the genome

Genes:
~1.2% coding
~2% non-coding

Regulatory regions:
~2%

Repetitive elements comprise another ~50% of the human genome
Information in the genome

Encyclopedia of DNA Elements: ENCODE
• 147 cell types / 1,640 data sets
• 80.4% of the human genome participates in at least one biochemical event
• 95% within 8 kb of a biochemical events
• 99% within 1.7 kb of a biochemical events

Clark et all 2015
• Capture sequencing / 24 cell types
• 22046 novel exons
• 10136 novel splice junctions

Reference human genome

• Human genomes vary significantly between individuals (~0.1%)

• Important things to note about the reference genome:
  – Is a composite sequence (i.e. does not correspond to anyone’s genome)
  – Is haploid (i.e. only 1 sequence)

• Computationally, a reference genome is used.
Reference human genome

• Genomic data is most common represented in two ways:

1. Sequence data – fasta format (.fa or .fasta)

```
>chr1
NNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNNACAGTACTGGCGGATTATAGGGAACACCCGGAGCATATGCTGTTTGTC
TCAGtagactcctaaatatgggattcctgggtttaaaagtaaaaaataaa
tatgttttaattttgtgaactgattaccatcagaattgtactgttctgtatccaccagcaatgtcttaggaatgcctgttttctccacaaagttttacttttt
....
```

2. Location data – bed format (.bed)

```
<table>
<thead>
<tr>
<th>chromosome</th>
<th>start</th>
<th>end</th>
<th>name</th>
<th>score</th>
<th>strand</th>
</tr>
</thead>
<tbody>
<tr>
<td>chr1</td>
<td>934343</td>
<td>935552</td>
<td>HES4</td>
<td>0</td>
<td>-</td>
</tr>
<tr>
<td>chr1</td>
<td>948846</td>
<td>949919</td>
<td>ISG15</td>
<td>0</td>
<td>+</td>
</tr>
</tbody>
</table>
```

All about genomic formats here - [http://genome.ucsc.edu/FAQ/FAQformat.html](http://genome.ucsc.edu/FAQ/FAQformat.html)
What we will cover

Structure of the human genome

Genomic information

- DNA (Sequence variation)
- RNA (Genes & gene expression)
- Regulation\Epigenetics
  - DNA methylation
  - Histone modification
  - Transcription factor binding
DNA: Sequence variation
Variations in DNA sequence

- **Cytological level:**
  - Entire chromosome (e.g. chromosome numbers)
  - Partial chromosome (e.g. segmental duplications, rearrangements, and deletions)

- **Sub-chromosomal level:**
  - Transposable elements
  - Short Deletions/Insertions, Tandem repeats

- **Sequence level:**
  - Single Nucleotide Polymorphisms (SNPs)
  - Small Nucleotide Insertions and Deletions (Indels; <=100bps)
Sequence variation

- **Single nucleotide polymorphisms (SNPs)**
  - DNA sequence variations that exist with members of a species.
  - They are inherited at birth and therefore present in all cells.

- **Somatic mutations**
  - Are somatic – i.e. only present in some cells.
  - Mutations are often observed in cancer cells.
Types of SNPs/Mutations

- Most SNPs and mutations fall in intergenic regions.
- Within genes, they can either fall in the non-coding or coding regions.
- Within coding regions, they can either not-change (synonymous) or change (non-synonymous) amino acids.
Effects of sequence variation

• Non-synonymous variants:
  – Missense (change protein structure)
  – Nonsense (truncates protein)

• Synonymous or non-coding variants:
  – Alter transcriptional/translational efficiency
  – Alter mRNA stability
  – Alter gene regulation (i.e. alter TF binding)
  – Alter RNA-regulation (i.e. affect miRNA binding)

Majority of sequence variation are neutral (<1% phenotype)
RNA: Genes and gene expression
A gene is a functional unit of DNA that is transcribed into RNA.

Total genes in the human genome – 57,445

Types of genes

- Protein coding genes, 20,318
- Pseudogenes, 14,181
- Long non-coding RNA, 13,562
- Small non-coding RNA, 8,998
- mRNA
- miRNA
- IncRNA

Source: GENCODE (version 18)
Protein coding genes

- ~ 20,000 in the human genome.
- Due to splicing one gene can make many proteins.
- Traditionally considered to be the most important functional unit of genomes.

Source: http://www.news-medical.net
MicroRNA (miRNA)

- Discovered in 1993.
- Plays a role in post-transcriptional regulation.
- Acts by either causing RNA degradation or inhibition of translation.
- Implicated in many aspects of health and disease including:
  - Development
  - Cancer
  - Heart disease
Long non-coding RNA (IncRNA)

- Recently described class of RNAs which often transcribed by PolIII promoters and often spliced.

- Unlike coding and miRNAs, IncRNA are less conserve.

- Non-coding transcripts > 200 nt in length.

- Many functions. Commonly recruitment of histone modifiers

Figure 4
Models of long noncoding RNA (IncRNA) mechanisms of action. (a) The IncRNAs can act as decoys that titrate away DNA-binding proteins, such as transcription factors. (b) These IncRNAs may act as scaffolds to bring two or more proteins into a complex or spatial proximity and (c) may also act as guides to recruit proteins, such as chromatin modification enzymes, to DNA; this may occur through RNA-DNA interactions or through RNA interaction with a DNA-binding protein. (d) Such IncRNA guidance can also be exerted through chromosome looping in an enhancer-like model, where looping defines the cis nature and spread of the IncRNA effect.
RNA expression

- Measuring the level of RNA in the sample.
- Generally microarray-, sequencing- or high-throughput PCR-based.
- Computation analysis and normalisation of expression data can be complicated.
RNA expression applications

- Relatively cheap and fast readout of the functional state of a cell
- Association with clinical features
  - sequence variations
  - response to therapy
  - patient survival
  ...
- Differential expression
  - between samples, or
  - between genes
RNA expression applications

- Differential expression of individual genes not necessarily informative.

- Genes are often grouped in gene-sets based on ontology or biological pathways.
Gene Regulation
Epigenetics
Epigenetics

• Mechanisms that alter cellular function independent to any changes in DNA sequence

• Mechanisms include:
  – Transcriptional regulation: Transcription Factors
  – Genome methylation
  – Histone modification / Nucleosome positioning
  – *Non-coding RNA*
Transcriptional regulation

- Transcription factors are proteins that bind DNA to co-regulate gene expression.
- Typically binds at gene promoters or enhancers.
DNA methylation

- DNA is methylated on cytosine's in CpG dinucleotides
Nucleosomes & Histones

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2. DNA is complexed with histones to form nucleosomes.
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Histone acetylation
NRC: nucleosome
HAT: histone ac
IBP: insulator box

UNSW
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Structure of the human genome

Genomic information
- DNA (Sequence variation)
- RNA (Genes & gene expression)
- Epigenetics
  - DNA methylation
  - Histone modification
  - Transcription factor binding

Data acquisition
- Microarrays
- Sequencing
- Chromatin IP
Array Technology

- Relies on fluorescence-based on hybridisation of DNA against complementary probe on array.

- Known molecule that can be converted to cDNA.
  - Expression array (probe for exonic DNA regions)
  - SNP array (probe for two alleles)
  - Methylation array (probe for bisulfide converted DNA)

- Limited by probes present on the array.

https://www.dkfz.de/gpcf/affymetrix_genechips.html
Array Technology

Images Processing
Quantification

Pre-processing
Backgrd. Subs., Norm.

https://www.dkfz.de/gpcf/affymetrix_genechips.html
Next-generation sequencing

Whole Genome Re-sequencing

Gene Expression

Targeted Re-sequencing

ChIP Sequencing

Other Applications

MicroRNA discovery
Next-generation sequencing (Illumina)
RNA-seq (vs mRNA Array)

- Alignment
  - human reference genome

- Quantification
  - mRNA/miRNA/IncRNA

- Statistics / Bioinformatics
Chromatin Immunoprecipitation Sequencing (ChIP-seq)

ChIP-seq of the seven transcription factors FLI1, ERG, GATA2, RUNX1, SCL, LYL1 and LMO

living cell

protein A

gene A

protein B

gene B

crosslink

lyse

fragment

precipitate

reverse crosslink

remove protein

DNA bound by protein A

High-throughput sequencing

Bioinformatics

ERG locus

Scale
chr21
Peak Calls

50 kb

FlI1

250

ERG

200

GATA2

1150

RUNX1

550

SCL

125

LYL1

650

LMO2

100

IgG

3K27Ac

mRNA

ERG

hg19

39,800,000

39,850,000
Pros/cons of each technology

• NGS
  – Greater dynamic range (only limited by depth of sequencing)
  – Coverage of genome does not need to be limited.
  – Many more applications from sequencing data.
  – Data analysis and management can be challenging.

• Microarrays
  – Microarrays are still significantly cheaper.
  – Largest public datasets are likely to be microarray based.
  – Data analysis pipelines are well standardised.
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UCSC Genome Browser

- Background
- Genome Assemblies
- Annotation Tracks
- Associated Tools
- Practical Exercise
About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. We encourage you to explore these sequences with our tools. The Genome Browser zooms and scrolls over chromosomes, showing the work genes that can be related in many ways. Blast quickly maps your sequence to the genome. The Table Browser provides convenient access to the data to examine expression patterns. Genome Graphs allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering. The team has a public mailing list where questions or feedback on the tools or data on this website can be posted. To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the UCSC Genome Browser team list.

News

24 October 2013 - Job Opening: UCSC Genome Browser Trainer

The Center for Biomolecular Science and Engineering (CBSE) at University of California Santa Cruz seeks an articulate, self-motivated educational in-person trainer for the UCSC Genome Browser at universities, hospitals, institutes, and professional meetings in the United States and international events. We are seeking experienced biologists and bioinformaticians who wish to use their knowledge of the Genome Browser to help users learn how to use the browser.

This position requires a Master’s degree in a biological science, a Ph.D. in molecular biology, experience in a research environment, working knowledge of teaching and training in a scientific environment. Preferred qualifications include a PhD in a relevant field, experience with video production, and teaching experience.

For more information and to apply for this position, see Job #1304619 on the UCSC Staff Employment website.

23 October 2013 - dbSNP Build 138 Available for hg19

We are pleased to announce the release of four tracks: derived from NCBI dbSNP Build 138 data, available on the human assembly (GRCh37) corresponding coloring and filtering options in the Genome Browser.

As was the case for the annotations based on the previous dbSNP build 137, there are four tracks in this release. One is a track containing all subtracks of this track and show interesting and easily defined subsets of dbSNP:

- Common SNPs (138): uniquely mapped variants that appear in at least 1% of the population or are 100% non-reference
- Flagged SNPs (138): uniquely mapped variants, excluding Common SNPs, that have been flagged by dbSNP as “clinically associated”
- Mult. SNPs (138): variants that have been mapped to more than one genomic location

By default, only the Common SNPs (138) are visible, other tracks must be made visible using the track controls.

You will find the four SNPs (138) tracks on the Human Feb. 2009 (GRCh37/hg19) browser in the “Variation and Repeats” group.

The tracks were produced at UCSC by Angie Hinrichs and Lurina Guravadoo. We'd like to thank the dbSNP group at NCBI for providing access.
Background
Background

Visualization of genomic data

- Graphical viewpoint on the very large amount of genomic sequence produced by the Human Genome Project.

Human Genome: 3,156,105,057 bp

- Focus turned from accumulating and assembling sequences to identifying and mapping functional landmarks

Genetic markers
Genes
SNPs
Points of regulation

- Visualization of Next-generation-sequencing data
Background

Client-side

Integrative Genomics Viewer*

- Application (Java) on the user’s machine
- Often difficult to install
- Does not have the extensive third-party data of the other browsers
- Much faster than web-based browsers

http://www.broadinstitute.org/igv/
Background

- Intronerator was developed by J. Kent to map the exon–intron structure of C. elegans RNAs mapped against genomic coordinates
Background

- Draft human genome sequence became available at the UCSC in 2000
- Intronerator was used as the graphics engine
UCSC Genome Browser
Genome Browser
http://genome.ucsc.edu/

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To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the Genome Browser E-mail List.

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The February 2009 human reference sequence (GRCh37) was produced by the Genome Reference Consortium. For more information about this assembly, see GRCh37 in the NCBI Assembly database.

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the User’s Guide for more information.

Request:  

<table>
<thead>
<tr>
<th>Request</th>
<th>Genome Browser Response:</th>
</tr>
</thead>
<tbody>
<tr>
<td>chr7</td>
<td>Displays all of chromosome 7</td>
</tr>
<tr>
<td>chrUn_gl1000212</td>
<td>Displays all of the unplaced contig gl1000212</td>
</tr>
<tr>
<td>20p13</td>
<td>Displays region for band p13 on chr 20</td>
</tr>
<tr>
<td>chr3:1-1000000</td>
<td>Displays first million bases of chr 3, counting from p-arm telomere</td>
</tr>
<tr>
<td>chr3:1000000+2000</td>
<td>Displays a region of chr3 that spans 2000 bases, starting with position 1000000</td>
</tr>
<tr>
<td>RH18061;RH80176</td>
<td>Displays region between genome landmarks, such as the STS markers RH18061 and RH80176, or chromosome bands 15q11 to 15q13, or SNPs rs1042522 and rs1800370. This syntax may also be used for other range queries, such as between uniquely determined ESTs, mRNAs, refSeqs, etc.</td>
</tr>
<tr>
<td>D16S3045</td>
<td>Displays region around STS marker D16S3045 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well.</td>
</tr>
<tr>
<td>AA205474</td>
<td>Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17</td>
</tr>
<tr>
<td>AC008101</td>
<td>Displays region of clone with GenBank accession AC008101</td>
</tr>
<tr>
<td>AF083811</td>
<td>Displays region of mRNA with GenBank accession number AF083811</td>
</tr>
<tr>
<td>PRNP</td>
<td>Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP</td>
</tr>
<tr>
<td>NM_017414</td>
<td>Displays the region of genome with RefSeq identifier NM_017414</td>
</tr>
<tr>
<td>NP_059110</td>
<td>Displays the region of genome with protein accession number NP_059110</td>
</tr>
</tbody>
</table>

pseudogene mRNA  | Lists transcribed pseudogenes, but not cDNAs                                           |
homeobox caudal  | Lists mRNAs for caudal homeobox genes                                                   |
zhc finger       | Lists many zinc finger mRNAs                                                          |
kruppel zinc finger | Lists only kruppel-like zinc fingers                                                |
huntington       | Lists candidate genes associated with Huntington’s disease                            |
zahler           | Lists mRNAs deposited by scientist named Zahler                                       |
Evans J.E.       | Lists mRNAs deposited by co-author J.E. Evans                                        |
Genome Assemblies

- Regular updates to genome assemblies to close gaps in genomic sequence, troubleshoot assembly problems and otherwise improve the genome assemblies.

- Shifting coordinates for known sequences and a potential for confusion and error among researchers, particularly when reading literature based on older versions.

- Frequently used assemblies hg18/hg19

- New assemblies increase genomic coverage 6-fold and have been deposited in GenBank.

- 127 genome assemblies have been released on 58 organisms (April 2012)
**Human (Homo sapiens) Genome Browser Gateway**

The UCSC Genome Browser was created by the Genome Bioinformatics Group of UC Santa Cruz. Software Copyright (c) The Regents of the University of California. All rights reserved.

### Sample position queries

A genome position can be specified by the accession number of a sequence or keywords from the GenBank description of an mRNA. The following list shows more information.

#### Request:

- **chr7**: Displays all of chromosome 7
- **chr8**: Displays all of the unplaced contig gll00212
- **chr3:1-1000000**: Displays region for band p13 on chr 20
- **chr3:1000000-2000**: Displays region of chr 3 that spans 2000 bases, starting from 1000000
- **RH10861**: Displays region between genome landmarks, such as rs1042522 and rs1800370. This syntax may mRNAs, refSeqs, etc.
- **D16S3106**: Displays region around STS marker D16S3106 from the Human Genome Project.
- **AC008113**: Displays region of EST with GenBank accession AC008113.
- **AF008113**: Displays region of mRNA with GenBank accession AF008113.
- **PRNP**: Displays region of genome with HUGO Gene Nomenclature.
- **NM_001741**: Displays the region of genome with RefSeq identifier NM_001741.
- **NP_059110**: Displays the region of genome with protein accession NP_059110.

**Genome Browser Response:**

- **chr2**: Displays all of chromosome 2
- **chrX**: Displays all of the unplaced contig gll00212
- **chr3:1-1000000**: Displays region for band p13 on chr 20
- **chr3:1000000-2000**: Displays region of chr 3 that spans 2000 bases, starting from 1000000

---

**RefSeq Genes**

<table>
<thead>
<tr>
<th>Gene Symbol</th>
<th>Chromosome</th>
<th>Position</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>FGFR2</td>
<td>chr10:123,227,429-123,343,066</td>
<td>FGR2</td>
<td>fibroblast growth factor receptor 2, transcript variant 2</td>
</tr>
</tbody>
</table>

**Non-Human RefSeq Genes**

<table>
<thead>
<tr>
<th>Gene Symbol</th>
<th>Chromosome</th>
<th>Position</th>
<th>Description</th>
</tr>
</thead>
<tbody>
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<td>FGR2</td>
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</tr>
</tbody>
</table>

---

**Basic Gene Annotation Set from ENCODE/GENCODE Version 17**

- **FGFR2**: fibroblast growth factor receptor 2, transcript variant 2.
chr10:123,237,844-123,353,481  115,638 bp.
Annotation tracks

Click on a feature for details.
Click or drag in the base position track to zoom in.
Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.

Use drop-down controls below and press refresh to alter tracks displayed. Tracks with lots of items will automatically be displayed in more compact modes.
Annotation tracks

- The database may contain any data that can be mapped to genomic coordinates and therefore can be displayed in the Genome Browser.

- Overview of tracks: [http://genome.ucsc.edu/cgi-bin/hgTracks](http://genome.ucsc.edu/cgi-bin/hgTracks)

- Three different categories:
  - computed at UCSC
  - computed elsewhere and displayed at UCSC
  - computed and hosted entirely elsewhere
Annotation tracks computed at UCSC

- Comparative genomic annotations as well as Convert and liftOver capabilities

- mRNAs and ESTs in GenBank are aligned to the reference assembly in separate tracks (75 million GenBank RNAs and ESTs, ~3 billion bases of the human reference assembly → 2 CPU-years of computing time)

- The Conservation composite track displays the results of the multiz algorithm that aligns the results from up to 46 pairwise Blastz alignments to the reference assembly (e.g. hg19 human assembly consumed 10 CPU-years)
Annotation tracks computed elsewhere and displayed at UCSC

Annotations that are not post-processed by the UCSC

- Probe sets for commercially available microarrays, copy-number variation from the Database of Genomic Variants or expression data from the GNF Expression Atlas
- Data Coordination Center for the ENCODE project allowing access to a large number of functional annotations in regards to gene regulation

Annotations that are post-processed by the UCSC

- dbSNP (Common SNPs, Flagged SNPs, Mult. SNPs)
- OMIM (OMIM Allelic Variant SNPs, OMIM Genes, OMIM Phenotypes)
Annotation tracks computed and hosted elsewhere

Data tracks are hosted remotely (no data stored at UCSC).
Tracks from the Epigenome project

[Image of a genome browser interface with various tracks and data visualization]
Associated Tools

- Tools other than the main graphic image account for 42% of traffic on the UCSC server
Sessions

Save Settings

Save current settings as named session:
- name: hg19
  - allow this session to be loaded by others

Save current settings to a local file:
- file: UCSC_Session.txt
  - file type returned: plain text
  - leave file blank to get output in browser window

Restore Settings

Use settings from another user's saved session:
- user: 
  - session name: 

Use settings from a local file:
- file: C:\Users\z3265235\Choose...

Use settings from a URL (http://..., ftp://...):

Sharing Sessions

There are several ways to share saved sessions with others.

- Each previously saved named session appears with Browser and Email links. Clicking on those links will load that session. The resulting Genome Browser page can be bookmarked.
- The Email link invokes your email tool with a message containing the Genome Browser page.
- If you have saved your settings to a local file, you can send email to others with a link to the file.
- If a saved settings file is available from a web server, you can send email to others with a link to the file.

Example links:
- genome.ucsc.edu/cgi-bin/hgSession
- http://...
Custom track

Add Custom Tracks

Display your own data as custom annotation tracks in the browser. Data must be formatted in BED, bigBed, bedGraph, GFF, GTF, WIG, bigWig, MAF, BAM, BED detail, Personal Genome SNP VCF, broadPeak, narrowPeak, or PSI formats. To configure the display, set track and browser fine attributes as described in the User’s Guide. Data in the bigBed, bigWig, BAM and VCF formats must be provided via a URL embedded in a track line in the box below. Publicly available custom tracks are listed here. Examples are here.

Paste URLs or data: Or upload: Choose Submit

Optional track documentation: Or upload: Choose

Click here for an HTML document template that may be used for Genome Browser track descriptions.
# Table Browser

## Using the Table Browser

This section provides brief line-by-line descriptions of the Table Browser controls. For more information on using the Table Browser, refer to the [User's Guide](#). To reset all user cart settings (including custom tracks), click [here](#).

<table>
<thead>
<tr>
<th>#</th>
<th>chr</th>
<th>chromStart</th>
<th>chromEnd</th>
<th>name</th>
<th>type</th>
<th>clinSign</th>
<th>phenotype</th>
<th>origin</th>
<th>otherIds</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>chr10</td>
<td>123247566</td>
<td>123247567</td>
<td>FGFR2</td>
<td>c.1882G</td>
<td>single nucleotid pathogenic</td>
<td>MedGen:C3281247, OMIM:614592,</td>
<td>germline</td>
<td>OMIM Allelic Variant:176943.0037</td>
</tr>
<tr>
<td>3</td>
<td>chr10</td>
<td>123256214</td>
<td>123256215</td>
<td>FGFR2</td>
<td>c.1694A</td>
<td>single nucleotid pathogenic</td>
<td>MedGen:C3281247, OMIM:614592,</td>
<td>germline</td>
<td>OMIM Allelic Variant:176943.0033</td>
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