Genomik Veri Tabanları

Doç. Dr. Deniz İlhan Topcu

İzmir Şehir Hastanesi

Tıbbi Biyokimya Bölümü

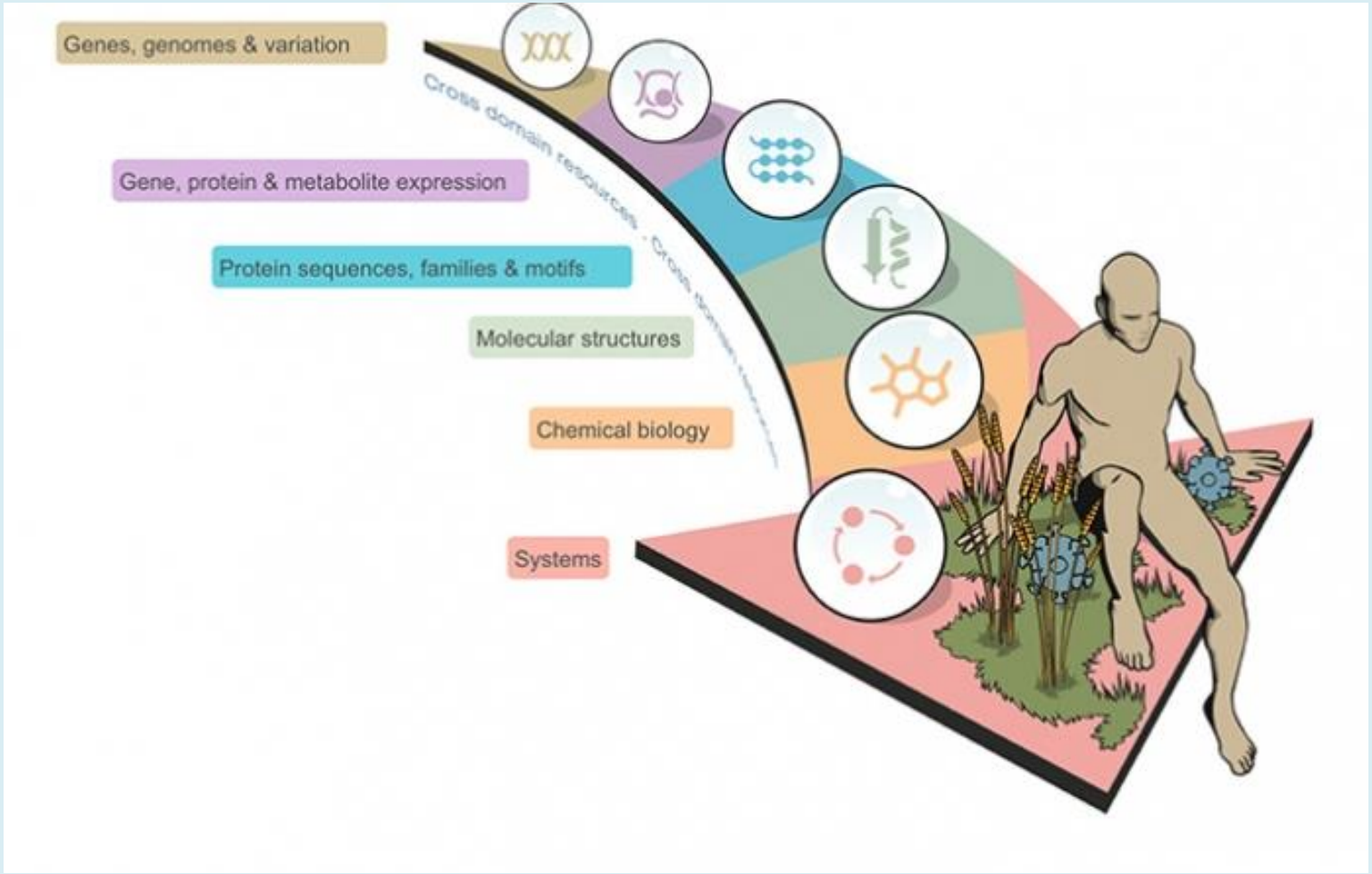
Kısa Özgeçmiş

- Tıp Doktoru (2004)
- Biyokimya Uzmanı (2009)
- Bilgisayar Mühendisi (2011)
- kullanıcısı (2014 - …)
- Tıbbi Biyokimya Doktora (2018)

Öğrenme Hedefleri

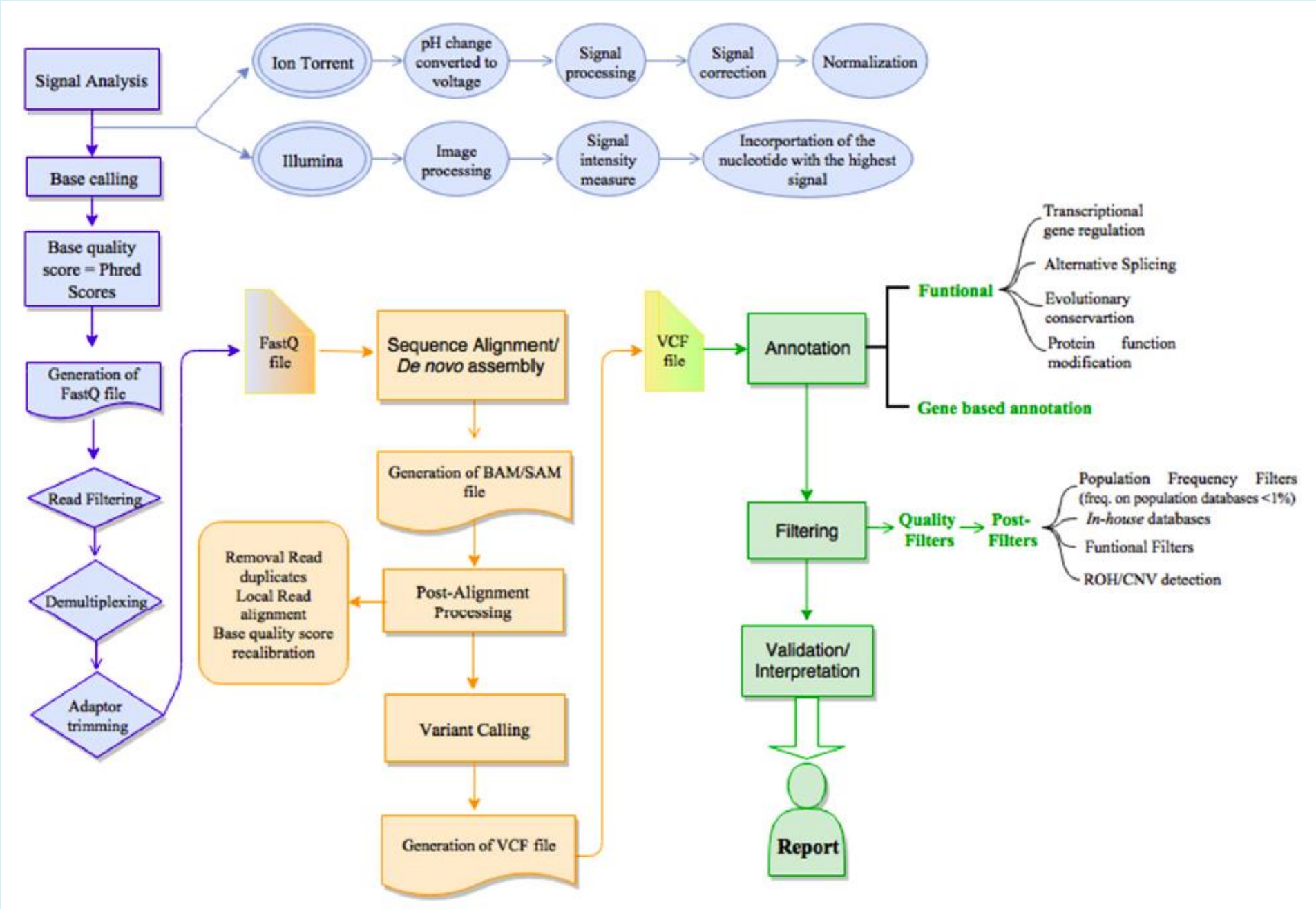
- Genomik veri tabanlarında bulunan farklı veri tipleri ve bunların biyoinformatikteki rolünü anlamak.
- Genomik veri tabanlarının temel işlevlerini ve türlerini öğrenmek.
- Transplantasyon immünolojisi kapsamında önemli genomik veri tabanlarını tanıyıp bu veri tabanlarından nasıl yararlanılacağını öğrenmek.
- Web tarayıcı üzerinden ve Bioconductor paketleri aracılığıyla genomik veri tabanlarından veri çekme becerilerini edinmek.

Biyoinformatik



https://www.ebi.ac.uk/training/online/courses/bioinformatics-terrified/what-bioinformatics/

Genomik Veri Türleri



Clinical Genetics. J Clin Med. 2020 Jan 3;9(1):132.

Genomik Veri Türleri

Sequence Data	Annotation
DNA	Nucleotide-level annotation
RNA	Protein-level annotation
Protein	Process-level annotation

Sekans Verisi için «FASTA» Dosyası

- FASTA dosya formatı, biyolojik sekans verilerini saklamak için en popüler formatlardan biridir.
- Bu metin tabanlı dosyalar,
  - Amino asit dizilerini (peptitler)
  - nükleotid dizilerini (DNA veya RNA) depolamak için kullanılabilir.
- Rutin olarak dizi açıklaması, veritabanı aramaları ve çoklu dizi hizalaması için kullanılırlar.
- İnsanlar ve bilgisayarlar tarafından anlaşılabilir
  - IUB/IUPAC amino asit kodlarını ve nükleik asit kodlarını kullanır
- ".fasta", ".fa", ".ffn", ".frn", ".fna", and ".faa"
- «>» ile başlar.

Nucleic Acids Research, Volume 38, Issue 6, 1 April 2010, https://doi.org/10.1093/nar/gkp1137

FASTA Dosyası Örneği

DNA

```
>NC_012920.1 Homo sapiens mitochondrion, complete genome
GATCACAGGTCTATCACCCTATTAACCACTCACGGGAGCTCTCCATGCATT
TGGTATTTTCGTCTGGGGGGTATGCACGCGATAGCATTGCGAGACGCTGG
AGCCGGAGCACCCTATGTCGCAGTATCTGTCTTTGATTCCTGCCTCATCCT
ATTATTTATCGCACCTACGTTCAATATTACAGGCGAACATACTTACTAAAG
TGTGTTA...
```

RNA

```
>Mus_musculus_tRNA-Ala-AGC-1-1 Ala (AGC)
GGGGGUGUAGCUCAGUGGUAGAGCGCGUGCUUAGCAUGCACGAGGCCCUGGGU
UCGAUCCCCAGCACCUCCA
```

Protein

```
>sp|P01210|PENK_HUMAN Proenkephalin-A OS=Homo sapiens OX=9606
GN=PENK
MARFLTLCTWLLLLGPGLLATVRAECSQDCATCSYRLVRPADINFLACVMECEG
KLPSLKIWETCKELLQLSKPELPQDGTSTLRENSKPEESHLLAKRYGGFMKRY
GGFMKKMDELYPMEPEEEANGSEILAKRYGGFMKKDAEEDDSLANSSDLLKE
LLETGDNRERSHHQDGSDNEE
```

FASTQ Dosya Türü

- FASTQ dosyaları FASTA'ya benzer ancak sekans verilerinin kalite skorunu da içerir.
- İki dosya arasındaki temel fark, FASTQ formatının ham sekanslama bilgilerini, özellikle de baz çağrılarıyla ilgili kalite puanlarını içermesidir.
- «@» ile başlar.

Nucleic Acids Research, Volume 38, Issue 6, 1 April 2010, https://doi.org/10.1093/nar/gkp1137

FASTQ Dosyası Örneği

```
1  @ERR000589.41 EAS139_45:5:1:2:111/1
2  CTTTCCTCCCTGCTTTCCTGGCCCCACCATTTCCAGGGAACATCTTGTCAT
3  +
4  3IIIIIIIIIIIII>1IIIFF9BG08E00I%IG+&?(4)%00646.C1#&(
5  @ERR000589.42 EAS139_45:5:1:2:1293/1
6  AGTTGTTAAAATCCAAGCCAATTAAGATAGTCTTATCTTTTTAAAAGAAAT
7  +
8  IIIIIGII.AIIII=?I9G-/II=+I=4?761BA2C9I+5A711+&>1$/I
```

Abuín Mosquera, José & Pichel, Juan & Pena, Tomas & Amigo, Jorge. (2016). SparkBWA: Speeding up the alignment of high-Throughput DNA sequencing data. PLOS ONE.

Anotasyon (Annotation)

GENOME ANNOTATION: FROM SEQUENCE TO BIOLOGY

Lincoln Stein

The genome sequence of an organism is an information resource unlike any that biologists have previously had access to. But the value of the genome is only as good as its annotation. It is the annotation that bridges the gap from the sequence to the biology of the organism. The aim of high-quality annotation is to identify the key features of the genome — in particular, the genes and their products. The tools and resources for annotation are developing rapidly, and the scientific community is becoming increasingly reliant on this information for all aspects of biological research.

Nat Rev Genet **2**, 493–503 (2001).

Anotasyon (Annotation)

- Gene locations
- Genetic variation
- Transcription factor binding sites
- Structural features of proteins

Anotasyon (Annotation)

- Tahmin edilen bir genin ürününün işlevinin tanımlanmasından oluşur (in-silico yaklaşımla).
- Bu tanımlamalar çeşitli biyoinformatik yazılımları aracılığıyla gerçekleştirilebilir:

  (1) Sinyal analizi (TATA box, start and stop kodonları, or poly-A sinyal tespiti)

  (2) İçerik analizi (for G 1 C content, codon usage, or dicodon frequency detection),

  (3) Benzerlik tespiti (benzer organizmalardaki proteinler, aynı organizmadaki mRNA referans genomları)

Nat Rev Genet **2**, 493–503 (2001).

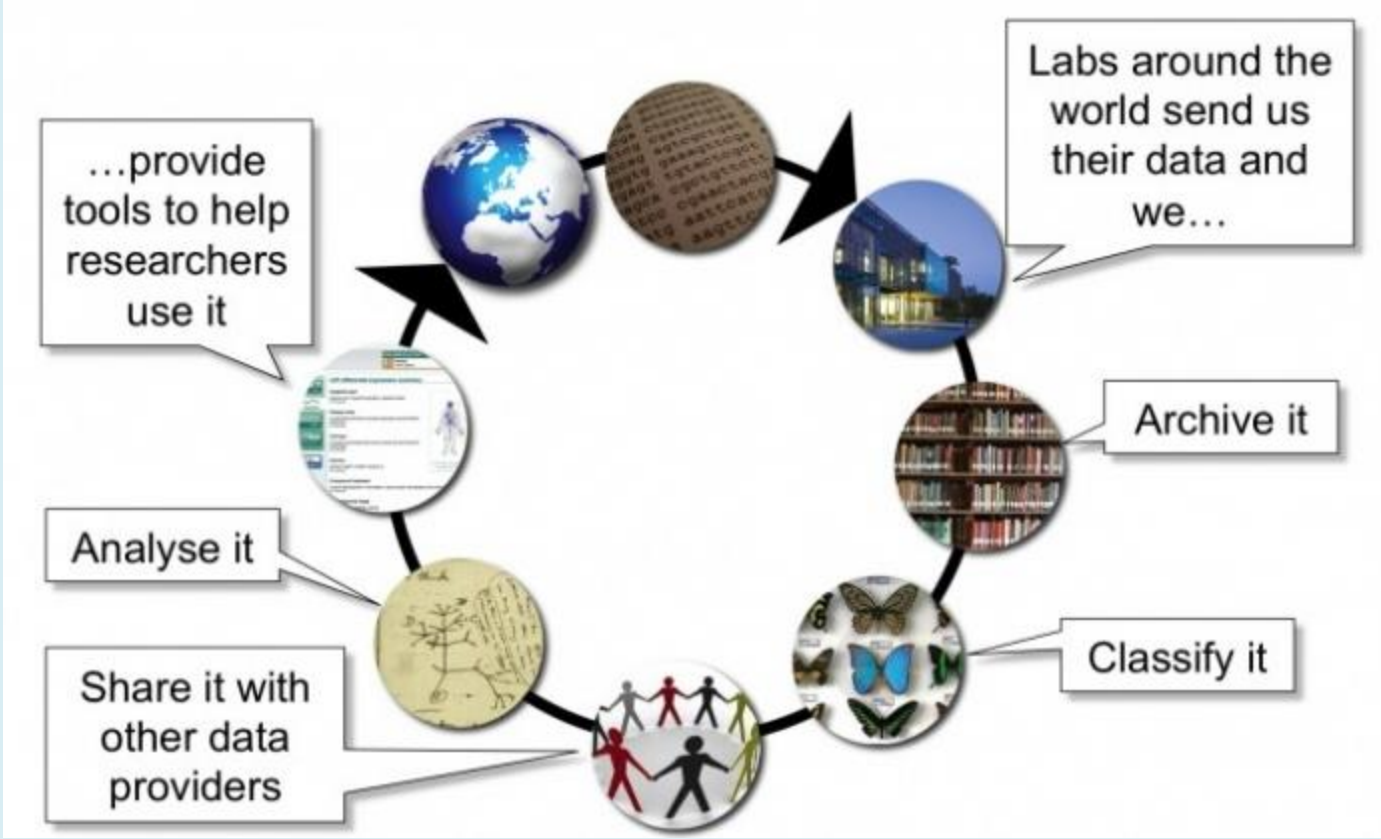
Anotasyon Örneği

Annotation

Chromosome	Start coordinates	End coordinates	Name (RefSeq ID)	Score	Strand
chr 1	9969621	10009136	NM_145692.2	0	-
chr 1	10024599	10038159	NM_013715.2	0	-
chr 1	10038217	10136768	NM_026493.3	0	+
chr 1	10046492	10136768	NM_001368816.1	0	+
chr 1	10137506	10232670	NM_001102430.1	0	-

Genomik Veri Tabanları

Genomik Veri Tabanları

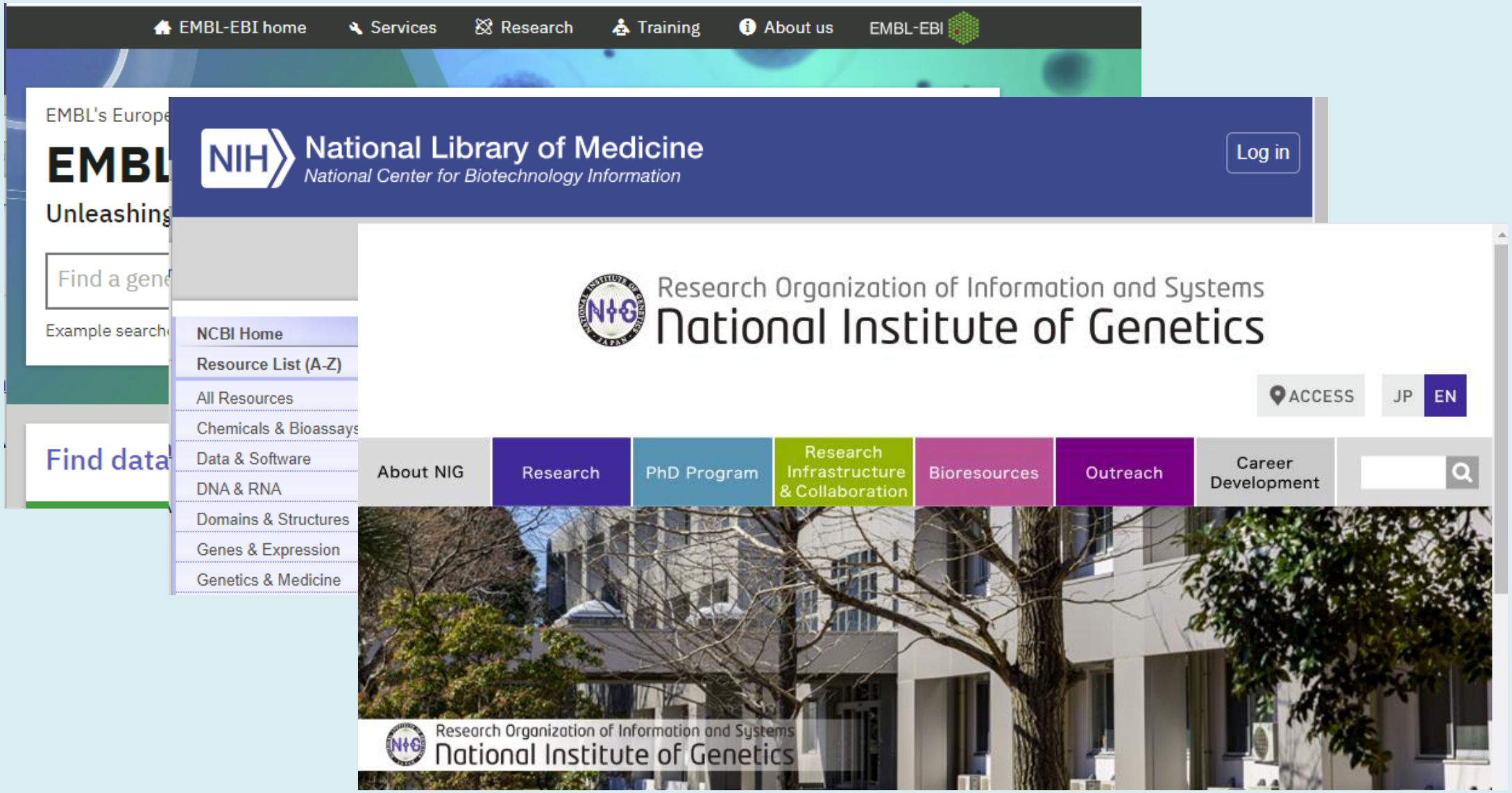


https://www.ebi.ac.uk/training/online/courses/bioinformatics-terrified/the-role-of-public-databases/

Genomik Veri Tabanlarının Kullanımı

- Bilgi deposu görevi görür.
  - Genomik verinin organize edilmesi ve düzenlenmesini sağlar.
- Araştırmacıların mevcut verileri incelemesine olanak sağlar.
- Veri türleri, bireyler ve organizmalar arasında verilerin paylaşılmasına ve karşılaştırılmasına olanak tanır.

Southwood D and Ranganathan S. Genome Databases and Browsers. In: Encyclopedia of Bioinformatics and Computational Biology Elsevier; 2019; pp. 251–256



EMBL-European Bioinformatics Institute (EMBL-EBI), National Center for Biotechnology Information, National Institute of Genetics of Japan

Primer ve Sekonder Veri Tabanları

	Primary database	Secondary database
Synonyms	Archival database	Curated database; knowledgebase
Source of data	Direct submission of experimentally-derived data from researchers	Results of analysis, literature research and interpretation, often of data in primary databases
Examples	ENA, GenBank and DDBJ (nucleotide sequence) ArrayExpress and GEO (functional genomics data) Protein Data Bank (PDB; coordinates of three-dimensional macromolecular structures)	InterPro (protein families, motifs and domains) UniProt Knowledgebase (sequence and functional information on proteins) Ensembl (variation, function, regulation and more layered onto whole genome sequences)

https://www.ebi.ac.uk/training/online/courses/bioinformatics-terrified/what-makes-a-good-bioinformatics-database/primary-and-secondary-databases/

Primer Veri Tabanları Örnekleri

- Genbank sequence database, NCBI
- Ensembl genome database, EMBL-EBI
- UCSC Genome Browser Database

NCBI: NCBI stands for the National Center for Biotechnology Information
EMBL-EBI:European Molecular Biology Laboratory's European Bioinformatics Institute
UCSC: University of California, Santa Cruz

National Library of Medicine
National Center for Biotechnology Information

Log in

All Databases | Search

All Databases
Assembly
Biocollections
BioProject
BioSample
Books
ClinVar
Conserved Domains
dbGaP
dbVar
Gene
Genome
GEO DataSets
GEO Profiles
GTR
Identical Protein Groups
MedGen
MeSH
NLM Catalog
Nucleotide

NCBI Home
Resource List (A-Z)
All Resources
Chemicals & Bioassays
Data & Software
DNA & RNA
Domains & Structures
Genes & Expression
Genetics & Medicine
Genomes & Maps
Homology
Literature
Proteins
Sequence Analysis
Taxonomy
Training & Tutorials
Variation

NCBI

...ter for Biotechnology Information advances science and health by providing access to ...enomic information.

...I | Mission | Organization | NCBI News & Blog

...bmit
...r manuscripts ...bases

Download
Transfer NCBI data to your computer

Learn
Find help documents, attend a class or watch a tutorial

Develop
Use NCBI APIs and code libraries to build applications

Analyze
Identify an NCBI tool for your data analysis task

Research
Explore NCBI research and collaborative projects

-> PubMed <-

Popular Resources
PubMed
Bookshelf
PubMed Central
BLAST
Nucleotide
Genome
SNP
Gene
Protein
PubChem

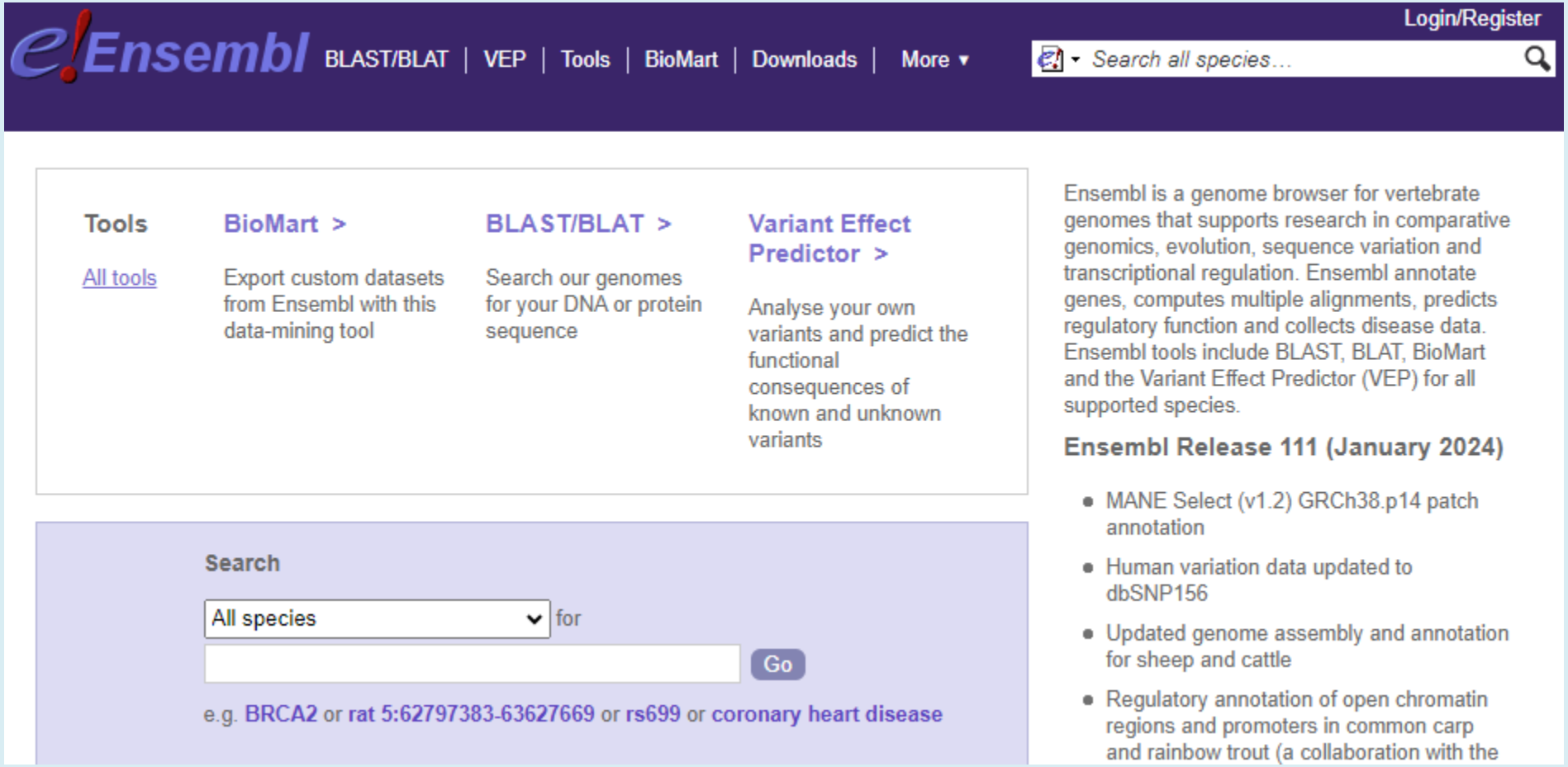
NCBI News & Blog

MedGen Users, We Want Your Feedback!
28 Mar 2024
Do you use NCBI's MedGen? If so, then you probably know it's NCBI's one-stop-

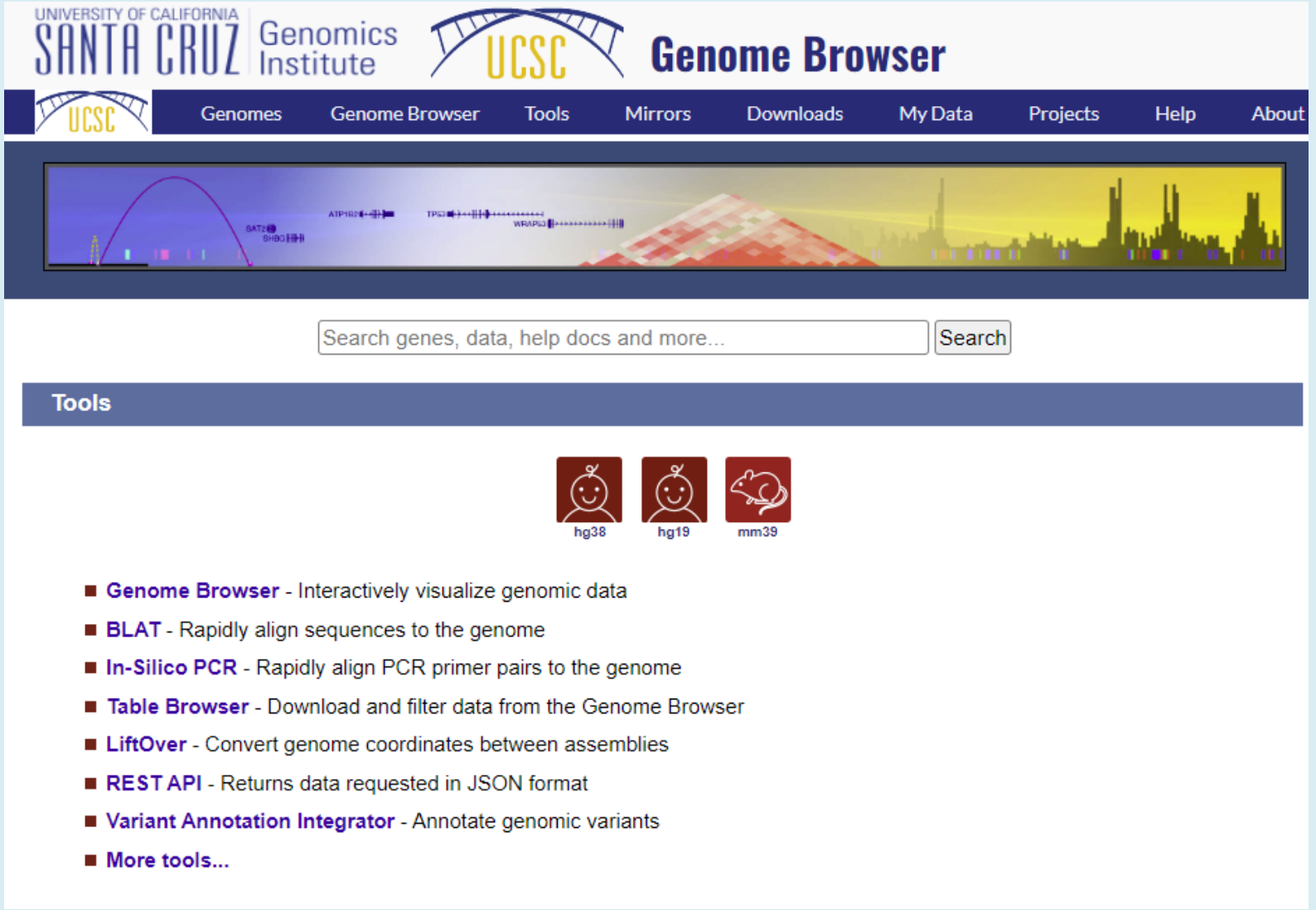
Changes to SRA Data Access on the Google Cloud Platform (GCP)
20 Mar 2024
Sequence Read Archive (SRA) data available via the Google Cloud Platform

Preview Upcoming Improvements to PubMed Central (PMC)
14 Mar 2024
Updated article display and journal list Since announcing the launch of a more

Ensembl Genome Database



UCSC Genome Browser Database



Sekonder Veri Tabanı Örnekleri

- clinVar

  https://www.ncbi.nlm.nih.gov/clinvar/

- COSMIC (Catalogue Of Somatic Mutations In Cancer)
  - https://cancer.sanger.ac.uk/cosmic
- CFTR2

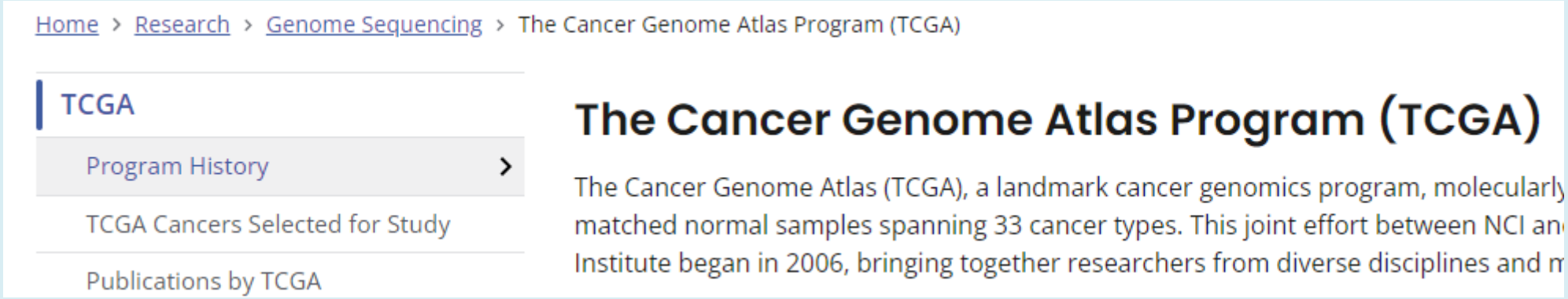
  https://cftr2.org/

- Human Hemoglobin Variants and Thalassemias (HbVar)

  https://globin.bx.psu.edu/hbvar/

Hibrid (Composite) Veri Tabanları

- Hem primer hem de sekonder genomik verileri bir araya getiren bir veritabanı türüdür.
- Ham DNA dizilimleri, RNA dizilimleri gibi doğrudan elde edilen verileri ve bu veriler üzerinde yapılan analizler sonucu elde edilen gen anotasyonları, protein etkileşimleri ve fonksiyonel bilgiler gibi işlenmiş verileri birlikte sunar
  - The Cancer Genome Atlas (TCGA)



Spesifik Veri Tabanları

- IPD-IMGT/HLA veri tabanı
- dbMHC
- Immune Epitope Database (IEDB)
- EPIMHC

IPD-IMGT/HLA Veri Tabanı

IPD-IMGT/HLA

Overview | IMGT/HLA | KIR | MHC | NHKIR | HPA | ESTDAB

IPD / IMGT/HLA

Welcome to IPD-IMGT/HLA

Release 3.55 (2024-01) Version Report - Build 9768104

The IPD-IMGT/HLA Database provides a specialist database for sequences of the human major histocompatibility complex (MHC) and includes the official sequences named by the WHO Nomenclature Committee For Factors of the HLA System. The IPD-IMGT/HLA Database was originally part of the international ImMunoGeneTics project (IMGT). For more information about the database and what data and tools are available please see our about page.

IPD-IMGT/HLA Veri Tabanı

- MHC ile ilişkili HLA genlerinin ayrıntılı genetik dizilerini içerir.
- Bu veri tabanı, transplantasyon immünolojisi ve otoimmün hastalıklar gibi alanlarda önemli olan HLA gen varyasyonlarını araştırmak için kullanılır.
- Dünya genelindeki popülasyonlarda HLA alel frekanslarını karşılaştırma imkanı sunar.
- İmmünoloji ve kişiselleştirilmiş tıp çalışmalarında referans bir kaynak olarak kullanılabilir.

IPD-IMGT/HLA Veri Tabanı

IPD-IMGT/HLA

Overview | IMGT/HLA | KIR | MHC | NHKIR | HPA | ESTDAB

IPD / IMGT/HLA / ALLELE LIST

Allele Query Tool

The IPD-IMGT/HLA Allele Query Tool provides advanced query interface allowing users to generate customizable queries to the IPD-IMGT/HLA database. This is available via a web interface or a POST API. Please see our help pages for documentation on the use of this tool. The default output displays all alleles and can be searched by typing a query in the box provided. More advanced searches can be performed by clicking the Advanced tab. The fields displayed in the output can also be changed by clicking the Fields tab.

dbMHC

- MHC ile ilgili DNA ve klinik verilerine açık erişim sağlayan bir platformdur.

- Bu verit abanı, dünya genelinde 90'dan fazla popülasyonda HLA alelleri ve haplotipleri gibi verilerin yanı sıra, kök hücre transplantasyonu gibi klinik veri setleri sunmaktadır.

dbMHC

NCBI Resources How To

⚠ **We are sorry, but the page you requested is**

dbMHC and IHWG data

The dbMHC database was an open, publicly accessible p
Complex (MHC). Data from IHWG workshops were provi

Data previously on the site are now available at ftp://ftp.n

If you have any specific questions,

0100010001100001011101000110000 1

IMMUNOGENOMICS DATA ANALYSIS WORKING GROUP

GCTAATGCACTGTACAGTATATCA

The immunogenomics data analysis working group (IDAWG) is an international collaboration of histocompatibility and immunogenetics investigators who share the goal of facilitating the sharing of immunogenomic data (HLA, KIR, etc.) and fostering the consistent analysis and interpretation of those data by the immunogenomics community and the larger genomics communities.

th International HLA and Immunogenetics
present its recommendations on topics of
W and Conference in 2012. IDAWG projects

Index of /pub/mhc/mhc/Final Archive

Name	Last modified	Size
Parent Directory		-
CN3D/	2018-02-02 13:18	-
Database/	2018-01-11 11:41	-
IHWG/	2018-01-11 11:48	-
Sequence_Viewer/	2018-01-09 12:08	-
alleles/	2018-01-09 12:09	-
dbMHC_FINAL_kit.xml.gz	2018-01-11 11:42	287K
dbMHC_FINAL_tube.xml.gz	2018-01-11 11:42	241K
dbMHC_Sequence_Alignment_Viewer.tar.gz	2018-01-11 11:42	4.0M

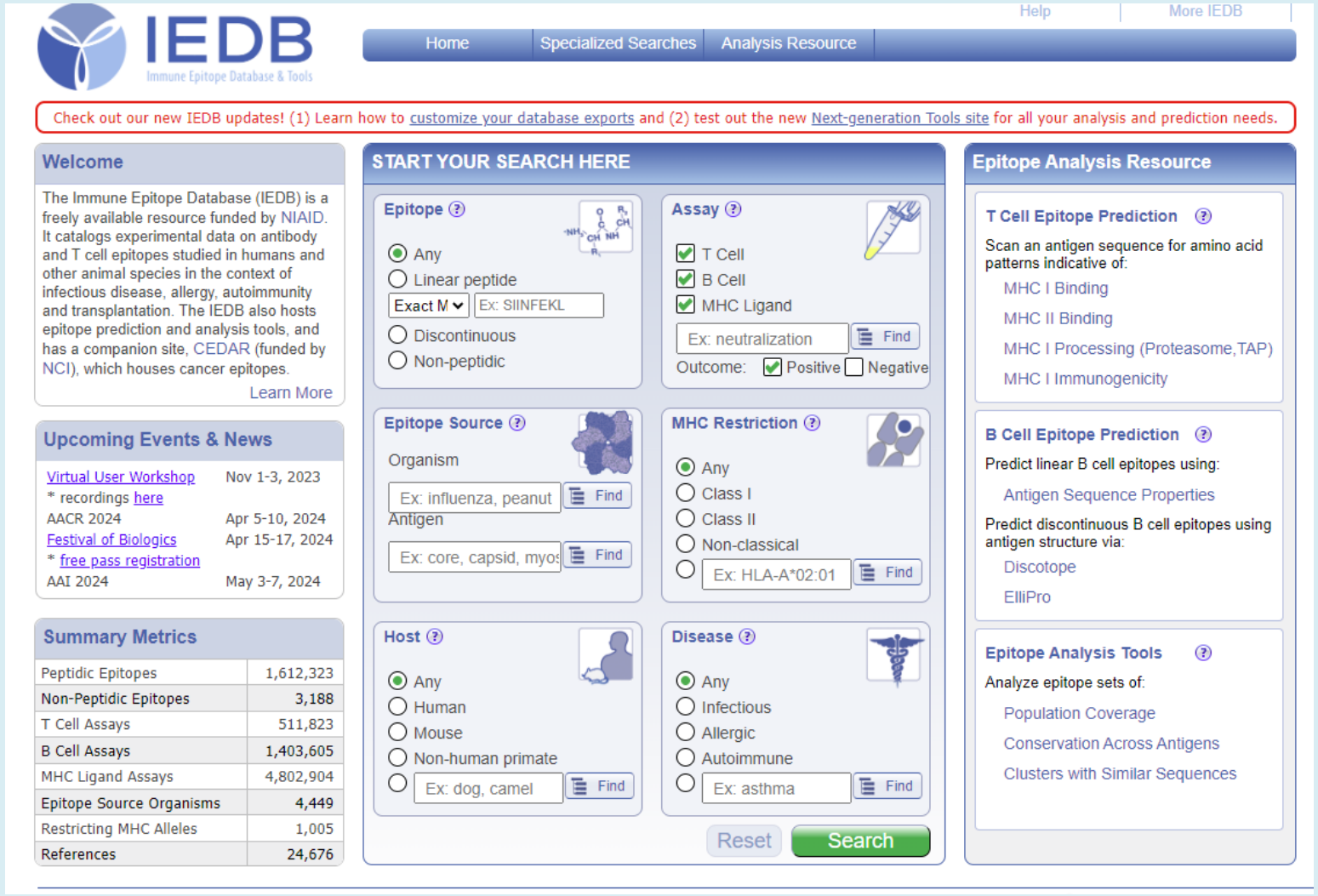
HHS Vulnerability Disclosure

Immune Epitope Database (IEDB)

- Immune Epitope Database (IEDB), enfeksiyon hastalıkları, alerjiler, otoimmün hastalıklar ve organ nakli gibi durumlarda antikor ve T hücresi epitoplarının kapsamlı verilerini içerir.
- İmmün tepkilerin ve aşı geliştirme süreçlerinin daha iyi anlaşılmasına yardımcı olacak epitop tahmin araçları sağlar.
- Veritabanı, epitopların yapısını, MHC ligand deneylerini ve T hücresi deney ayrıntılarını içerir.

www.iedb.org ; www.niaid.nih.gov

Immune Epitope Database (IEDB)



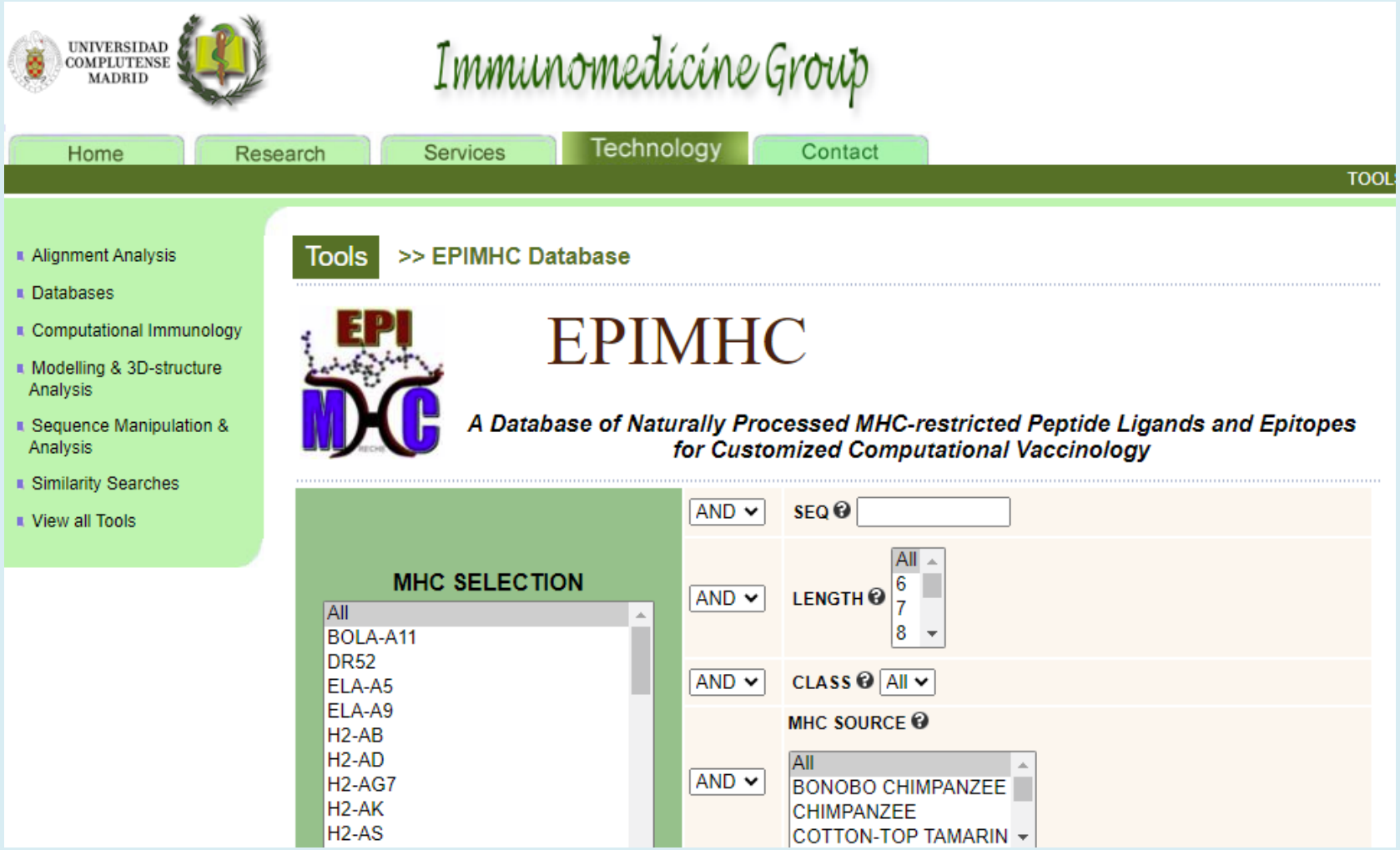
www.iedb.org ; www.niaid.nih.gov

EPIMHC

- EPIMHC, proteinlerde gözlemlenen MHC-binding peptidler ve T hücresi epitoplarını içeren ilişkisel bir veri tabanıdır.
- Veri tabanı, 84 tümör ilişkili antijen dahil olmak üzere çeşitli kaynaklardan 4867 farklı peptid sekansına ait bilgi içerir.
- Aşı geliştirme ve özel immünolojik araştırmalar için çeşitli araçlar sağlar.

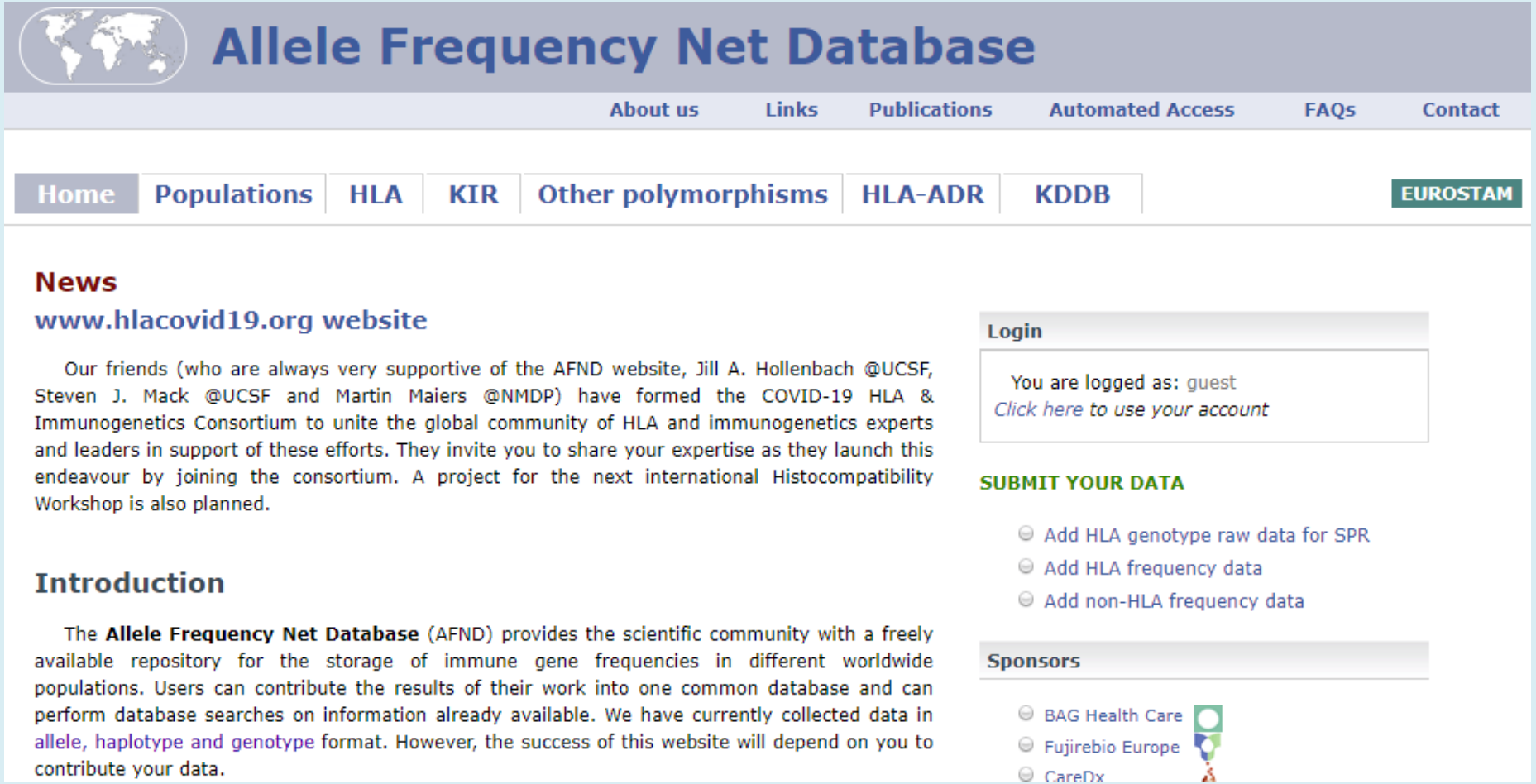
bio.med.ucm.es/epimhc/

EPIMHC



bio.med.ucm.es/epimhc/

Diğer Veri Tabanları



https://www.allelefrequencies.net/

Genomik Veri Tabanlarına Nasıl Ulaşılır?

Genomik Veri Tabanlarına Nasıl Ulaşılır?

- Web Browser
- REST API
- Bioconductor

Web Browser ile Ulaşım Örneği

- https://iastate.pressbooks.pub/molecularplantbreeding/chapter/introduction-to-bioinformatics/

REST API

Ensembl REST API Endpoints

Archive

Resource	Description
GET archive/id/:id	Uses the given identifier to return its latest version
POST archive/id	Retrieve the latest version for a set of identifiers

Comparative Genomics

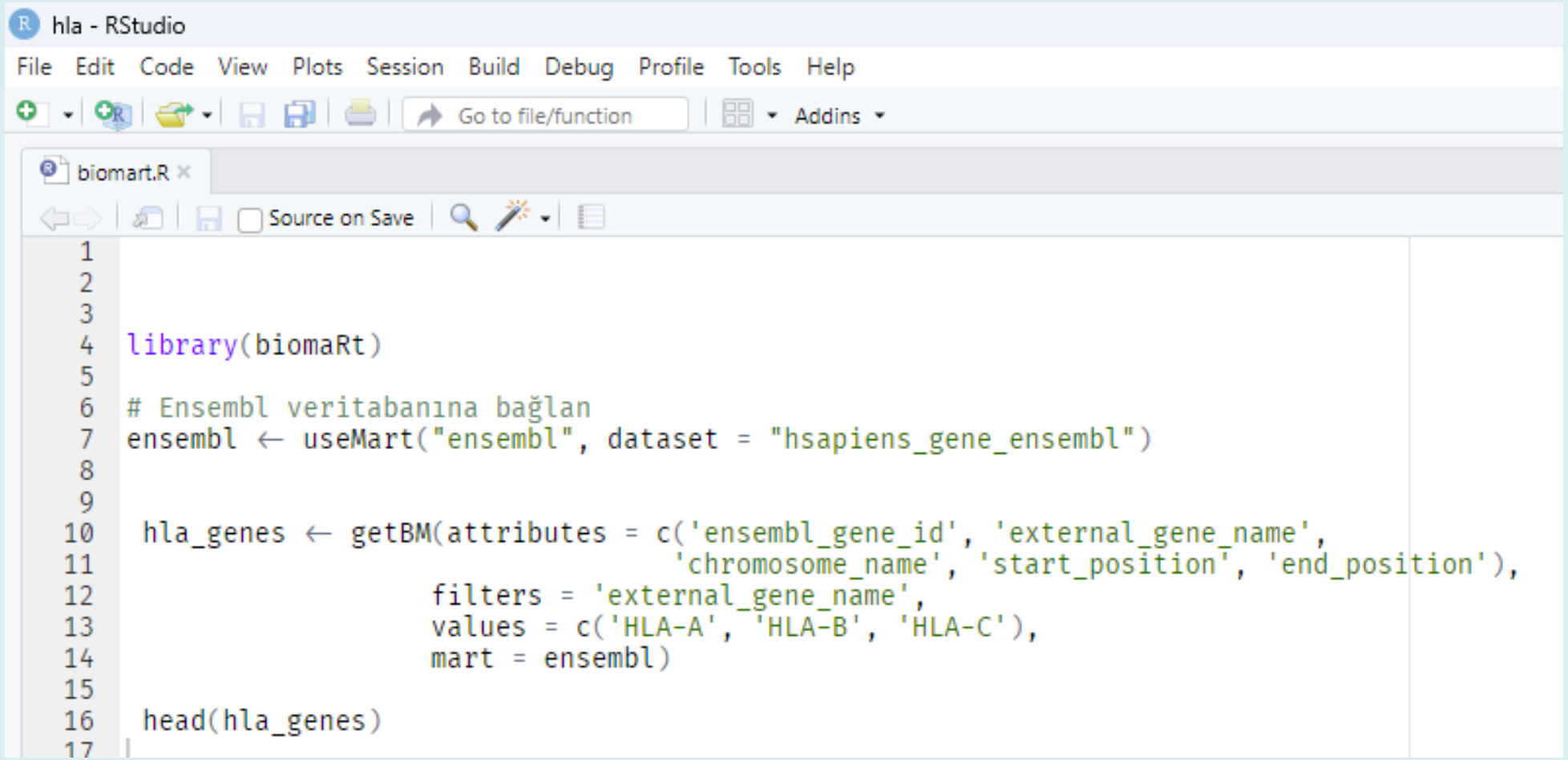
Resource	Description
GET cafe/genetree/id/:id	Retrieves a cafe tree of the gene tree using the gene tree stable identifier
GET cafe/genetree/member/id/:id	Retrieves the cafe tree of the gene tree that contains the gene / transcript / translation stable identifier. DEPRECATED: use cafe/genetree/member/id/:species/:id instead.
GET cafe/genetree/member/symbol/:species/:symbol	Retrieves the cafe tree of the gene tree that contains the gene identified by a symbol
GET cafe/genetree/member/id/:species/:id	Retrieves the cafe tree of the gene tree that contains the gene / transcript / translation stable identifier in the given species
GET genetree/id/:id	Retrieves a gene tree for a gene tree stable identifier
GET genetree/member/id/:id	Retrieves the gene tree that contains the gene / transcript / translation stable identifier. DEPRECATED: use genetree/member/id/:species/:id instead.

https://rest.ensembl.org/

Bioconductor

- Veri tabanlarına erişim için Bioconductor kütüphaneleri
  - **BiomaRt** Ensembl and other databases
  - **Ensembldb** Ensembl database
  - **GenomicFeatures** UCSC Genome Browser
  - **ipdDb** PD-IMGT/HLA Database
  - **GEOquery** Gene Expression Omnibus (GEO)
  - **TxDb.Hsapiens.UCSC.hg19..**
  - **AnnotationHub**
  - **AnnotationDbi**

Bioconductor Örneği: BiomaRt

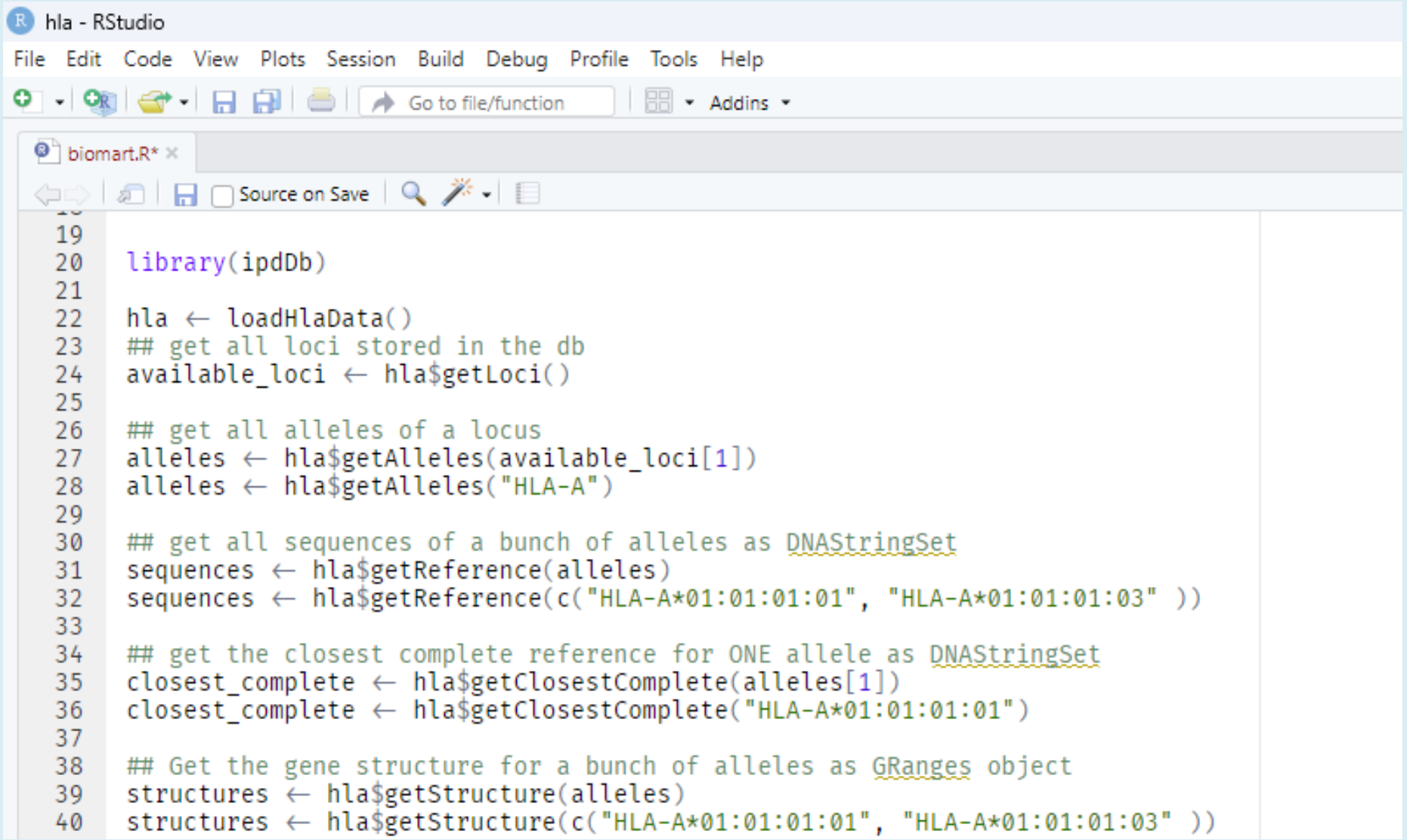



```r
library(biomaRt)

# Ensembl veritabanına bağlan
ensembl ← useMart("ensembl", dataset = "hsapiens_gene_ensembl")


 hla_genes ← getBM(attributes = c('ensembl_gene_id', 'external_gene_name',
                                  'chromosome_name', 'start_position', 'end_position'),
                   filters = 'external_gene_name',
                   values = c('HLA-A', 'HLA-B', 'HLA-C'),
                   mart = ensembl)

 head(hla_genes)
```

Bioconductor Örneği: BiomaRt

```
>
> # Ensembl veritabanına bağlan
> ensembl ← useMart("ensembl", dataset = "hsapiens_gene_ensembl")
>  hla_genes ← getBM(attributes = c('ensembl_gene_id', 'external_gene_name',
+                                  'chromosome_name', 'start_position', 'end_position'),
+                   filters = 'external_gene_name',
+                   values = c('HLA-A', 'HLA-B', 'HLA-C'),
+                   mart = ensembl)
>  head(hla_genes)
  ensembl_gene_id external_gene_name      chromosome_name start_position end_position
1 ENSG00000231834               HLA-A HSCHR6_MHC_MANN_CTG1         1195801      1200442
2 ENSG00000224320               HLA-A  HSCHR6_MHC_COX_CTG1         1369022      1489345
3 ENSG00000235657               HLA-A  HSCHR6_MHC_DBB_CTG1         1144955      1265390
4 ENSG00000224608               HLA-B HSCHR6_MHC_MANN_CTG1         2578541      2665857
5 ENSG00000237022               HLA-C HSCHR6_MHC_MANN_CTG1         2577801      2581172
6 ENSG00000228299               HLA-C  HSCHR6_MHC_DBB_CTG1         2526549      2529920
>
```

Bioconductor Örneği: ipdDb



Bioconductor Örneği: ipdDb

```
> closest_complete
DNAStringSet object of length 1:
    width seq                                                                                                            names
[1]  3503 CAGGAGCAGAGGGGTCAGGGCGAAGTCCCAGGGCCCCAGGCGTGGCTCTCAGGGTCTCAGGC...TCTCCATCTCTGTCTCAACTTCATGGTGCACTGAGCTGTAACTTCTTCCTTCCCTATTAAAA HLA-A*01:01:01:01
> closest_complete
DNAStringSet object of length 1:
    width seq                                                                                                            names
[1]  3503 CAGGAGCAGAGGGGTCAGGGCGAAGTCCCAGGGCCCCAGGCGTGGCTCTCAGGGTCTCAGGC...TCTCCATCTCTGTCTCAACTTCATGGTGCACTGAGCTGTAACTTCTTCCTTCCCTATTAAAA HLA-A*01:01:01:01
> structures
GRanges object with 34 ranges and 4 metadata columns:
                             seqnames    ranges strand |        type        name  complete     frame
                                <Rle> <IRanges>  <Rle> | <character> <character> <logical> <integer>
  HLA-A*01:01:01:01 HLA-A*01:01:01:01     1-300      + |         UTR      5' UTR      TRUE         0
  HLA-A*01:01:01:01 HLA-A*01:01:01:01   301-373      + |        Exon      Exon 1      TRUE         1
  HLA-A*01:01:01:01 HLA-A*01:01:01:01   374-503      + |      Intron    Intron 1      TRUE         0
  HLA-A*01:01:01:01 HLA-A*01:01:01:01   504-773      + |        Exon      Exon 2      TRUE         3
  HLA-A*01:01:01:01 HLA-A*01:01:01:01  774-1014      + |      Intron    Intron 2      TRUE         0
                ...               ...       ...    ... .         ...         ...       ...       ...
  HLA-A*01:01:01:03 HLA-A*01:01:01:03 2840-2981      + |      Intron    Intron 6      TRUE         0
  HLA-A*01:01:01:03 HLA-A*01:01:01:03 2982-3029      + |        Exon      Exon 7      TRUE         3
  HLA-A*01:01:01:03 HLA-A*01:01:01:03 3030-3198      + |      Intron    Intron 7      TRUE         0
  HLA-A*01:01:01:03 HLA-A*01:01:01:03 3199-3203      + |        Exon      Exon 8      TRUE         3
  HLA-A*01:01:01:03 HLA-A*01:01:01:03 3204-3300      + |         UTR      3' UTR      TRUE         0
  -------
  seqinfo: 2 sequences from an unspecified genome; no seqlengths
```

Diğer İlişkili R Kütüphaneleri

- BIGDAWG
- HLAminer
- immunoClust
- ComplexHeatmap

Bridging ImmunoGenomic Data Analysis Workflow Gaps (BIGDAWG)

BIGDAWG

Derek Pappas, Ph.D. (dpappas@chori.org)

2021-10-23

Overview

'Bridging ImmunoGenomic Data-Analysis Workflow Gaps' ('BIGDAWG') is an integrated analysis system that automates the manual data-manipulation and trafficking steps (the gaps in an analysis workflow) normally required for analyses of highly polymorphic genetic systems (e.g., the immunological human leukocyte antigen (HLA) and killer-cell Immunoglobulin-like receptor (KIR) genes) and their respective genomic data (immunogenomic) (Pappas DJ, Marin W, Hollenbach JA, Mack SJ. 2016. 'Bridging ImmunoGenomic Data Analysis Workflow Gaps (BIGDAWG): An integrated case-control analysis pipeline.' Human Immunology. 77:283-287). Starting with unambiguous genotype data for case-control groups, 'BIGDAWG' performs tests of Hardy-Weinberg equilibrium, and carries out case-control association analyses for haplotypes, individual loci, specific HLA exons, and HLA amino acid positions.

https://cran.r-project.org/web/packages/BIGDAWG/vignettes/BIGDAWG.html

HLAminer

HLAminer

HLAminer

HLAminer is a software for HLA predictions from next-generation shotgun (NGS) sequence read data and supports direct read alignment and targeted *de novo* assembly of sequence reads.

It has been used successfully to predict expressed HLA genes in the large TCGA (The Cancer Genome Atlas) cancer cohort.

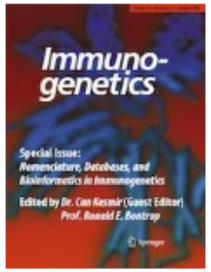
HLAminer uses TASR as its main assembly engine, which in turn is derived from SSAKE, the first *de novo* genome assembler published. A manuscript describing the methodology is published in the peer-reviewed journal *Genome Medicine*.

https://www.bcgsc.ca/resources/software/hlaminer

Sonuç ve Öneriler

- Genomik veri tabanları, transplantasyon immünolojisi araştırmalarında kritik öneme sahiptir.
- Buradaki bilgiler uygun donör eşleşmelerini ve greft reddi mekanizmalarını anlamada kullanılabilir.
- Veri tabanlarına ulaşmada günümüzdeki bir çok farklı araç bulunmaktadır.
- Bioconductor gibi araçlar, özel veri analizi için bilimsel çalışmaları desteklemektedir.

Kaynaklar - 1



Volume 72, Issue 1–2

February 2020

Special Issue: Nomenclature, Databases, and Bioinformatics in Immunogenetics

Issue Editors: Can Kesmir, Ronald E. Bontrop

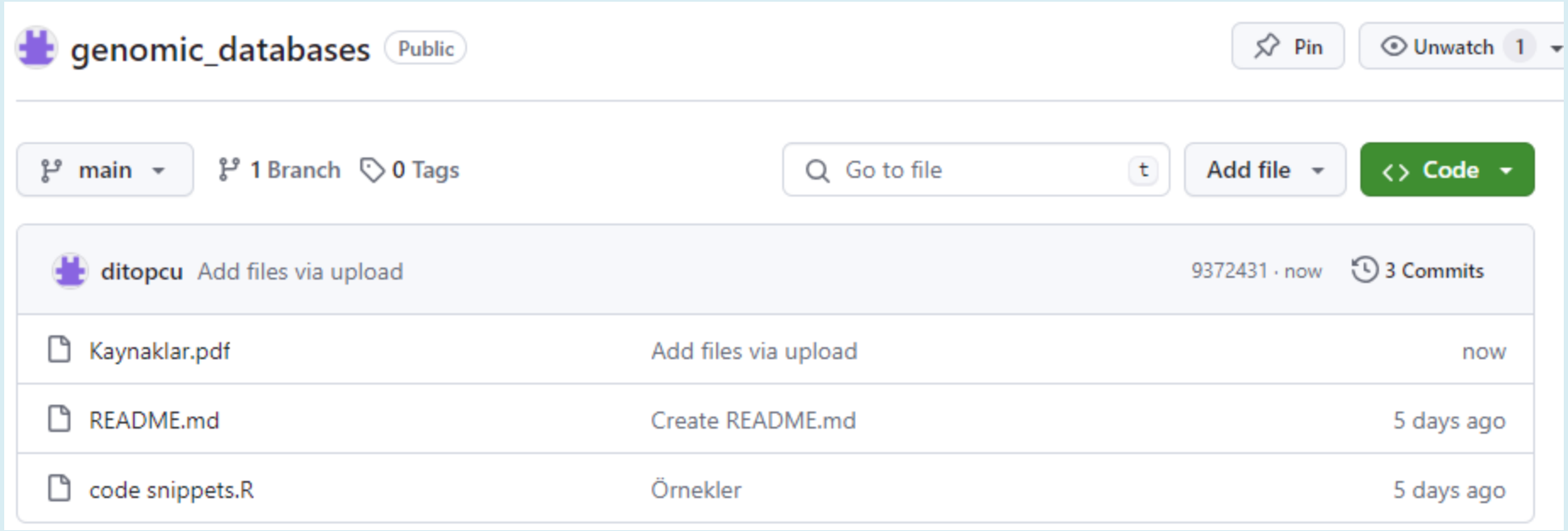
Review

Bioinformatics and Computational Tools for Next-Generation Sequencing Analysis in Clinical Genetics

Rute Pereira [1,2,*], Jorge Oliveira [2,3,†] and Mário Sousa [1,2,†]

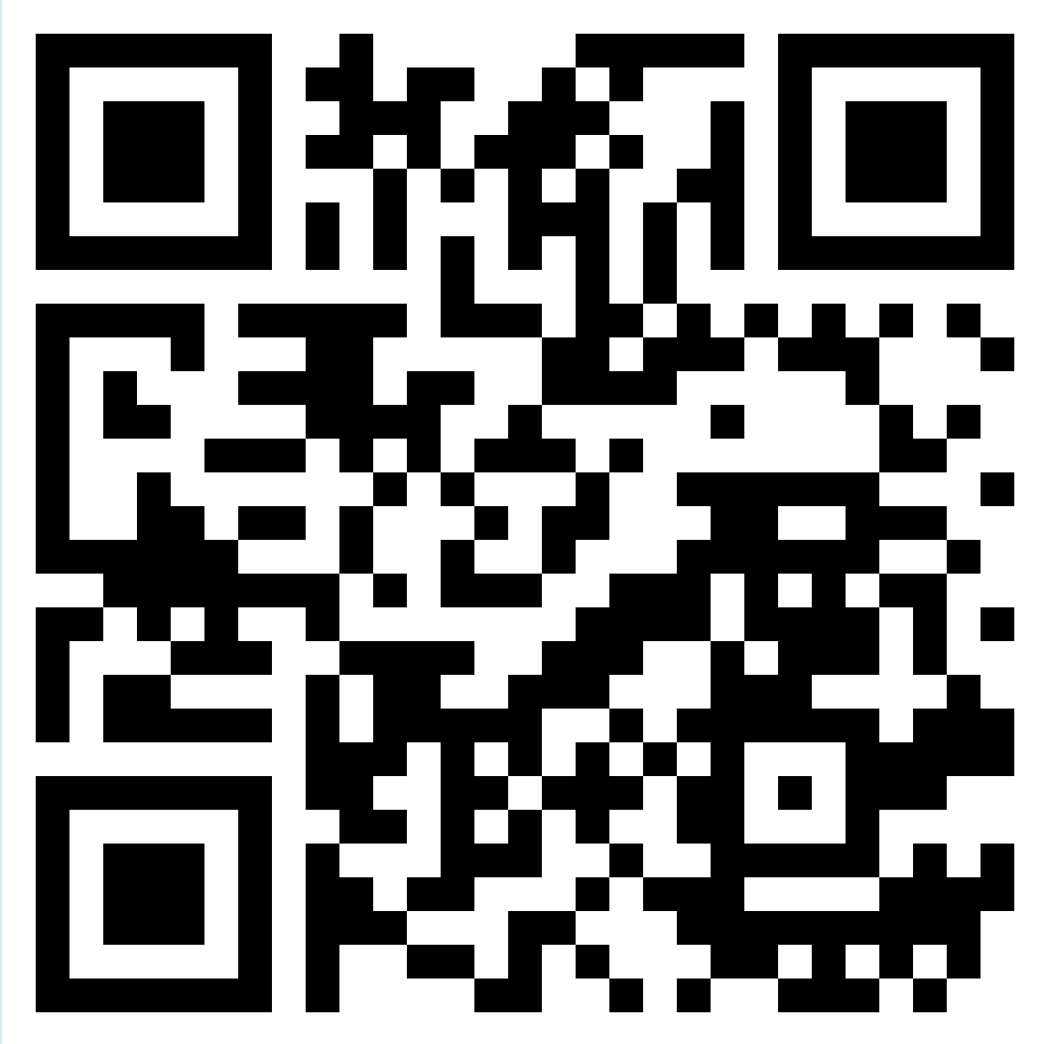
https://link.springer.com/journal/251/volumes-and-issues/72-1, J Clin Med. 2020 Jan 3;9(1):132.

Kaynaklar - 2



https://github.com/ditopcu/genomic_databases

Kaynaklar - 2



https://github.com/ditopcu/genomic_databases

Teşekkürler !

Soru ?

Katkı +