cancercelllines.org - a Cancer Cell Line Variant Knowledge Resource Built on the Beacon Framework

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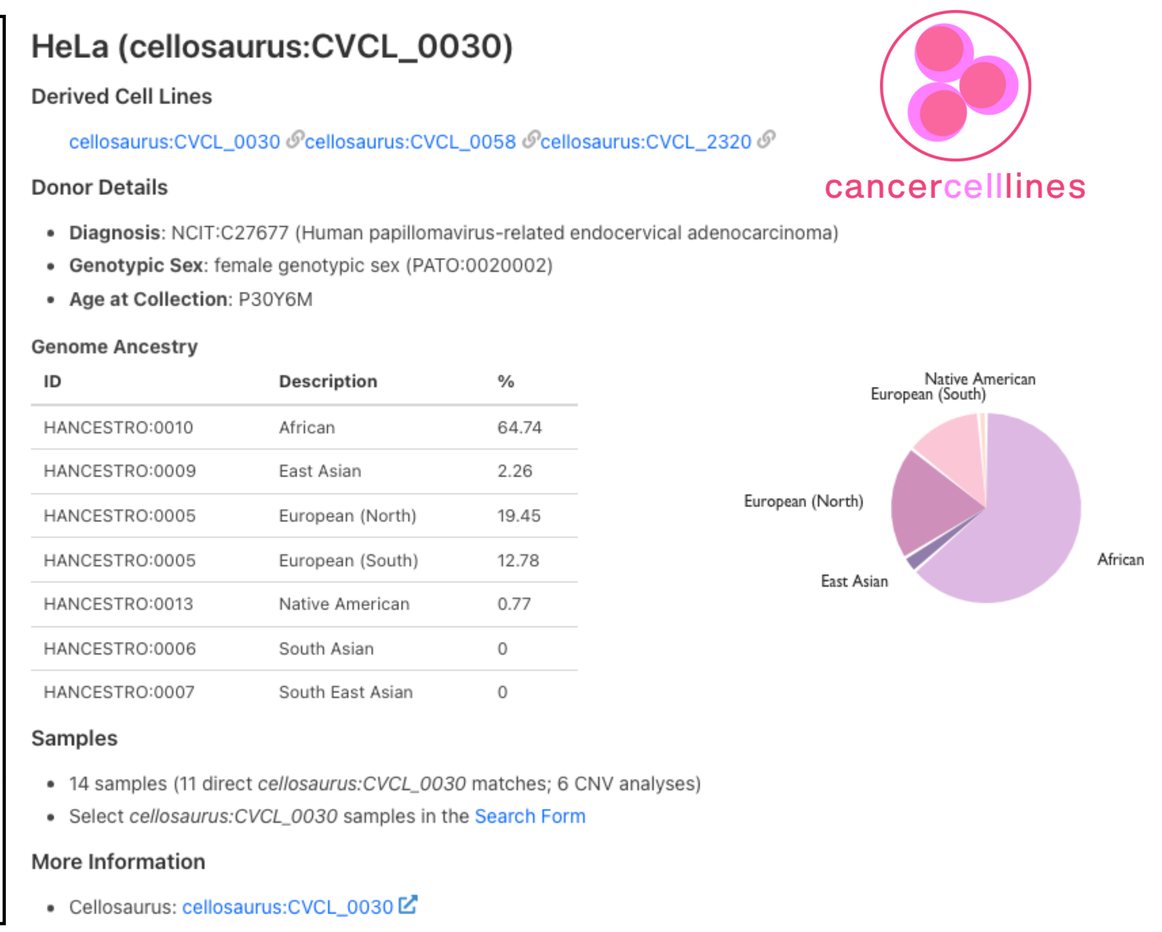
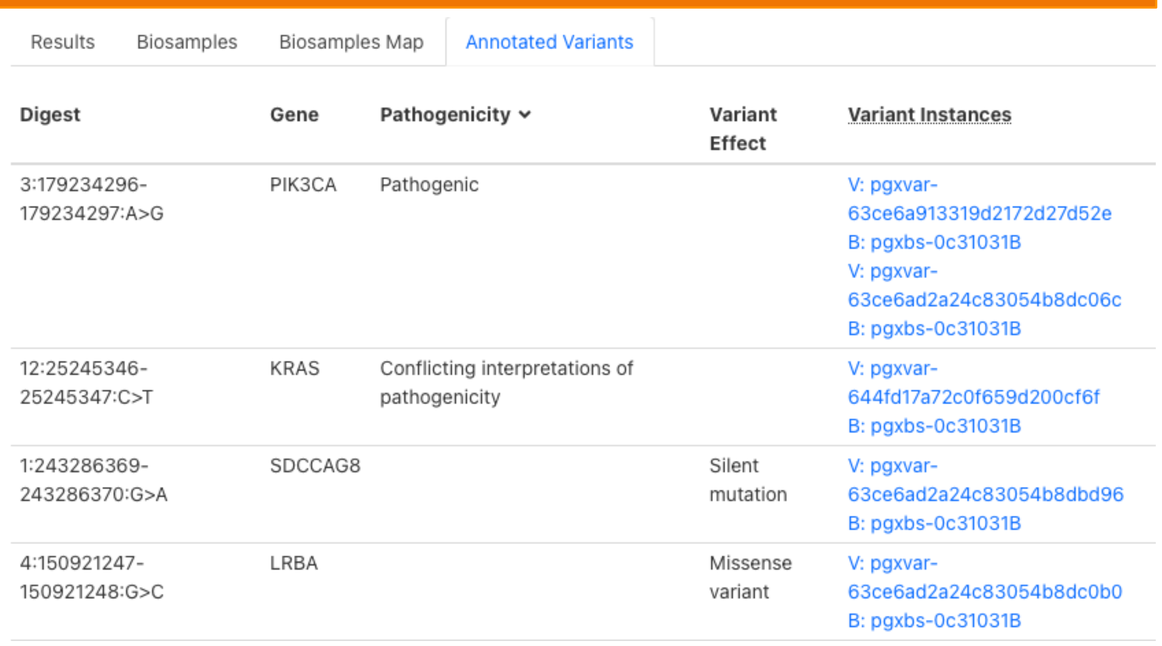
- The Beacon framework was created for global and federated queries. To facilitate variant sharing, we have built our novel cancer cell line resource on the Beacon framework.
- cancercelllines.org is the daughter of progenetix (a cancer copy number variant (CNV) database), including over 5600 cell line CNVs
- We have mapped additional known cancer cell line variants from resources like CCLE and ClinVar to our database, resulting in 16178 cell lines total from 400 different NCIT disease classifications.
- Known information about each cell line (from Cellosaurus) is displayed on the cell line page *e.g.* age at collection, genome ancestry and genotypic sex.
- Each cell line is represented hierarchically and includes an NCIT diagnostic code. Both hierarchies can be accessed in the database.



Annotated Single Nucleotide Variants

- We mapped known cancer cell line single nucleotide variants (SNVs), including parental SNVs annotated on Cellosaurus.
- SNVs mapped from ClinVar show known severity of the variant and disease ontologies.
- SNVs mapped from CCLE include data about variant effect.
- As an example, some of the annotated SNVs for breast cancer cell line MDA-MB-453 are listed on the right.



Copy Number Profiles of Cancer Cell Lines

- Over 5600 unique copy number profiles from more than 2000 cancer cell lines.
- Copy number profiles from cancer cell lines and their origins (from Progenetix) can be used for comparison.
- On the right are 4 CNV profiles for tumor and its cell line example.
- Fun fact: cell line MDA-MB-435 has been found to be misidentified. It was thought to originate from breast carcinoma but has been shown to originate from amelanotic melanoma instead.

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