NGS Catalog Manual

8/14/2012

1. Data Source

Species	NGS Technologies	No. Of Records	No. of Journal	Data collected from
Human	Exome Sequencing, Whole Genome Sequencing, Transcriptome sequencing (RNA-Seq)	285	64	Literature

2. Description of Column Headings for NGS Catalog

We have collected the following information about NGS studies,

Paper Information:

Author: Last name of first author
Date: Publication data (online (electronic publication) data if available)
Journal: Full journal name
Title: Title of paper (linked to PubMed abstract)

Study Information:

Sequencing Technology: Next-generation sequencing method used in the study, include exome sequencing, RNA sequencing and whole genome sequencing
Platform: Sequencing platform used in study
Disease/Trait: Disease or trait examined in study
Mutation type: Somatic mutation or germline mutation examined in study
Population: Population of the samples in study
Samples: Sample constitution in study
Max Read Length (bp): Maximum read length in study
Average Fold Coverage: The average coverage of sequencing data in study
Mapping Tool(s): Alignment tools used in study
Variant Calling Tool(s): Variant calling tools used in study
Reference Genome: Reference genome used in study
Public SNPs Database: Known SNPs used in study

Results Summary:

SNVs: Number of total and novel SNVs detected
Short Indels: Number of total and novel indels detected

Copy Number Variations: Number of total and novel copy number variations
Large Structural Variants: Number of total and novel structural variations
Reported gene(s): Gene(s) reported by author
Identified gene fusion(s): Identified gene fusion by author
Identified mutation(s):

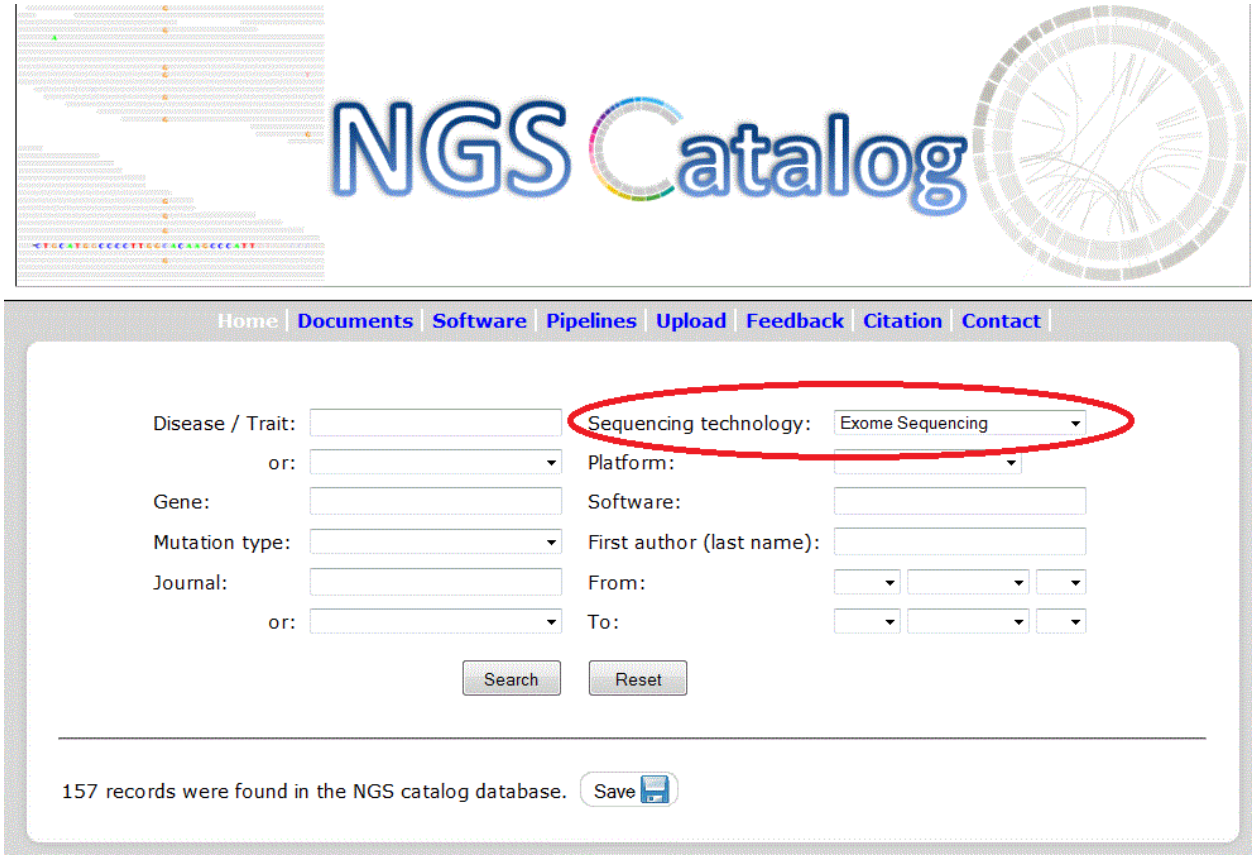
3. Web interface

Users can browse or search the data at different levels.
Browse:
1) browse by sequencing technology;
2) browse by platform;
3) browse by disease;
4) browse by publication date (beginning date and end date);
5) browse by journal.

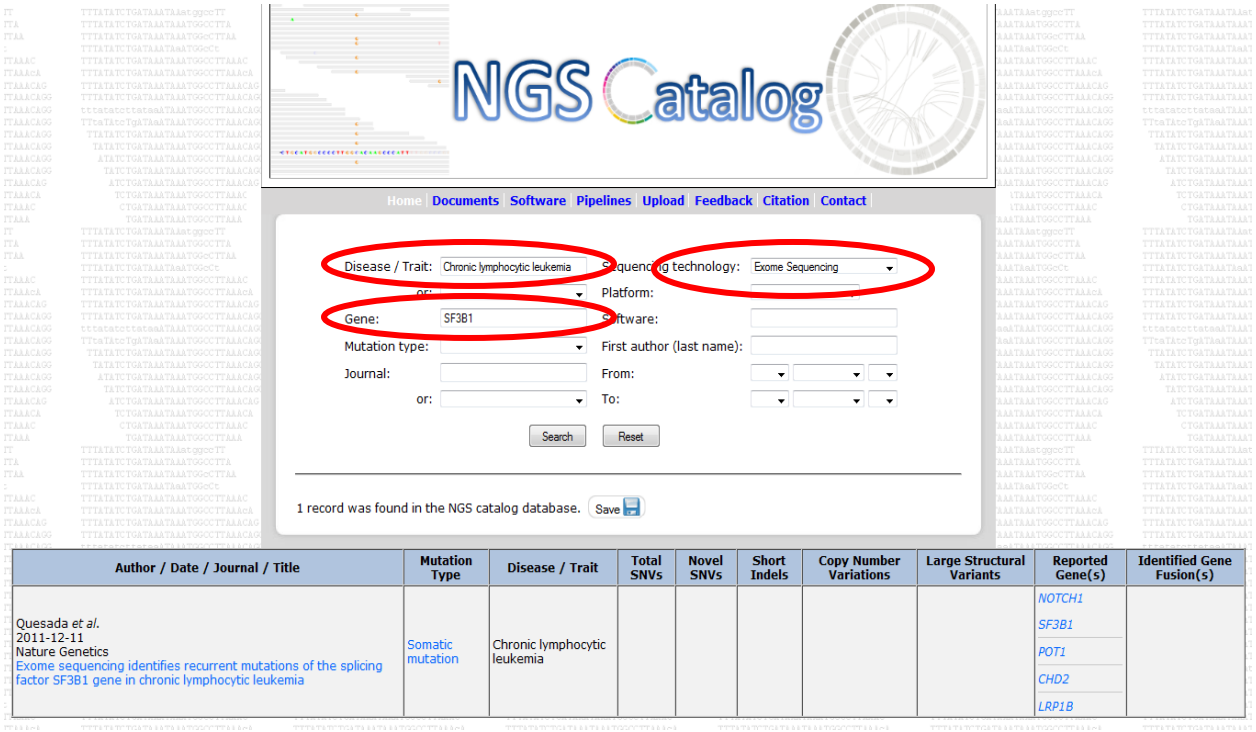
Search:
1) Quick search can be performed by choose any from "***Sequencing technology***", "***Platform*** ", "***Disease/Traits***", "***Journal***" and ***Date of journal published*** (begining date and end date) or performed by entering value in the search box of ***Software names*** (e.g., BWA), ***Author name*** (i.e last name of the first author, like Puente), and ***Gene names*** (e.g., MYD88).

2) If searching without any input, it will display all the results from our database.

Searching based on browse options:



Searching based on multiple selections:



User can get more detailed information like the sample information, information about the tools used (mapping and variant calling tools), PubMed link to the paper and NCBI gene link to the genes etc. by clicking on the particular paper title in the results table. In addition, we have provided the mutations information by clicking on mutation type (somatic mutation or germline mutation)

4. Data Download:

Users can download the results option using the **SAVE** option above the results tables. You can either download the file in CSV format.