N_xClinical™ Sample Querying Example Sheet

Case, spacing, and use of quotation marks are important so please make note of that in the examples below. For instance, all quotation marks must be in the ASCII format, otherwise the query will not work. Type quotation marks directly into the query field rather than copying and pasting from other documents as often text editing tools, such as WORD, use smart quotes rather than ASCII quotes.

Querying by Sample Name *Enter the entire or part of the sample name; "*" can be used as a wildcard*	
Search for all samples beginning with "Batch"	• Batch*
Search for samples that have "Batch" and the word "male" somewhere in the name	• *Batch*male*
Search for samples with the number "80021"	• *80021*
Search for all samples ending with "June2019"	• *June2019
Querying by Sample Attribute *Sample attributes must be preceded by "Factor:"; If a factor value is to be specified, it is entered with "="; Multi-term factor names (e.g. "Start Date") need to be in quotes.*	
Search for Male samples	• Factor:Gender=Male
Search for sample where the Attribute and/or value is a multi-word term.	• Factor: "Reported Condition"= "short stature"
Searching for all samples belonging to one family	• Factor: "Linked Sample Id"=Adams
Querying by Quality Score *Returns samples based on specified score range. Value must be in quotes.*	
Searching for samples with quality score less than or equal	• dna_attribute:quality<= "0.10"
Benign/pathogenic classified CN events overlapping a region *Returns samples with classified copy number events overlapping the specified bp location/gene. The only classification values supported are "benign" and "pathogenic"; they are case sensitive. Both regions and gene symbols are supported. Only works for CN events.*	
Benign or pathogenic CN events overlapping a region	• pathogenic:chr13:46573687-51607314
Benign or pathogenic CN events overlapping a gene	• pathogenic:PTEN

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Classified events and event types *Returns samples with specified event type having the specified classification. Any user-defined classification value can be specified. Event types are categorized as CN change, allelic event, and sequence variant.*	
CN change (CN Gain, CN Loss, High Copy Gain, Homozygous Copy Loss) classified as "Likely Pathogenic"	• sample_term: "DNAData:cn_cls_sum:Likely Pathogenic"
Allelic event (AOH, Allelic Imbalance) classified as "Benign"	• sample_term: "DNAData:snp_cls_sum:Benign"
Sequence Variant event (SNV, Deletion, Insertion) classified as "SV in Dominant Gene"	• sample_term: "DNAData:SeqVar:cls_sum:SV in Dominant Gene"
Event queries *Returns samples with specified events overlapping specific bp location/genes/cytoband. Events must be in lowercase; gene symbols can be in lowercase or uppercase. Events that can be queried: gain, loss, aoh, seqvar*	
Events overlapping a gene	• gain:PTEN • aoh:grb2
Events overlapping a region	• aoh:chr11:30507990-83574562 • loss: chr11:1-200000000 • seqvar: chr7:91724344-91724344
Events on a single chromosome (use 1 as start bp and bp equal to or longer than chromosome length for end)	• loss: chr11:1-200000000
Events at a cytoband	• loss:1q31.3
Queries using conditional (AND/OR) *A + before the query term indicates that the sample must meet the condition (AND); term without a preceding symbol indicates an OR statement; a – before the query term indicates that the sample must not meet the condition*	
Searching for sample with either a gain OR loss overlapping EGFR	• loss:EGFR gain:EGFR
Searching for samples with a loss overlapping PTEN AND a gain overlapping EGFR	• +loss:PTEN +gain:EGFR
Searching for samples that must contain a gain of EGFR and not a loss of FOXA1	• +gain:EGFR -loss:FOXA1

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Querying by processing details *Returns samples based on selected processing details. Note: when searching based on the processing date, the software searches based on when processing ended, not when it started.*	
Searching for samples processed within a certain timeframe: +dna_attribute:proc_ended_timestamp>=”2016-09-10” +dna_attribute:proc_ended_timestamp<”2017-01-07”	• +dna_attribute:proc_ended_timestamp>=”2016-09-10” • +dna_attribute:proc_ended_timestamp<”2017-01-07”
Searching for samples processed by a specific user	• dna_attribute:proc_by=”admin”
Querying by phenotypes *Returns samples containing selected phenotypes; phenotypes must be entered as HPO IDs, not text. Use * before and after the ID to indicate that any other HPO ID can be present to include samples that may contain more than the specified phenotype. If looking for samples containing only a single phenotype, don’t use * before or after.*	
Searching for samples that contain the phenotype “Seizures”	• factor:Phenotypes=”*HP:0001250*”
Searching for samples that contain the phenotype “Seizures” or “Global Developmental Delay”	• factor:Phenotypes=”*HP:0001263*”
Searching for samples that contain only the phenotype “Seizures” and no other phenotypes	• factor:Phenotypes=”HP:0001250”

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