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Breaking the Data Analysis Bottleneck: Solutions That Work For RNA and Exome Sequencing

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ABRF
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“It’s just a little research, it shouldn’t be a problem?”

The challenges of collaborative research

- Communication
- Flow of information / data sharing
- Patient confidentiality

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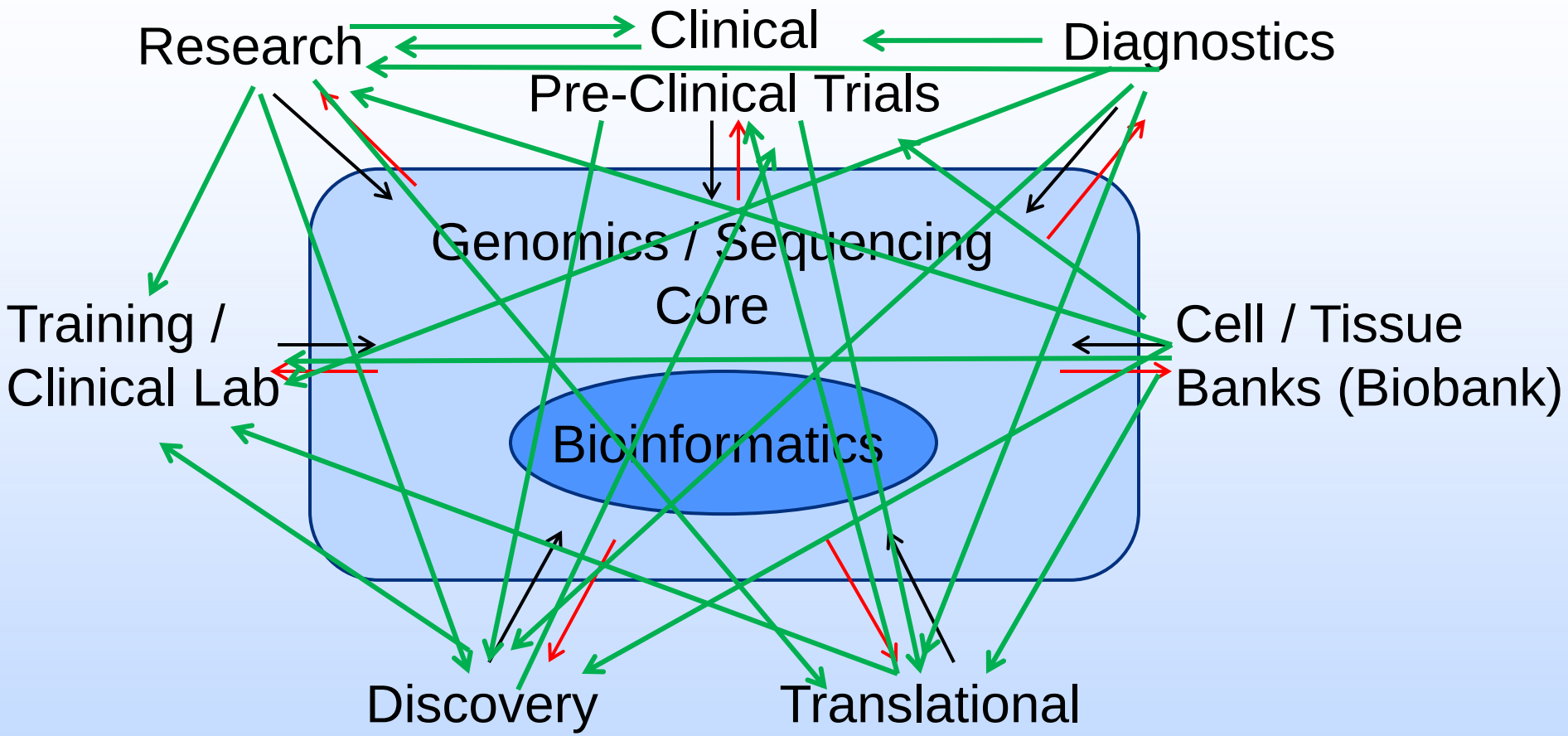
Clinicians



Laboratory Research

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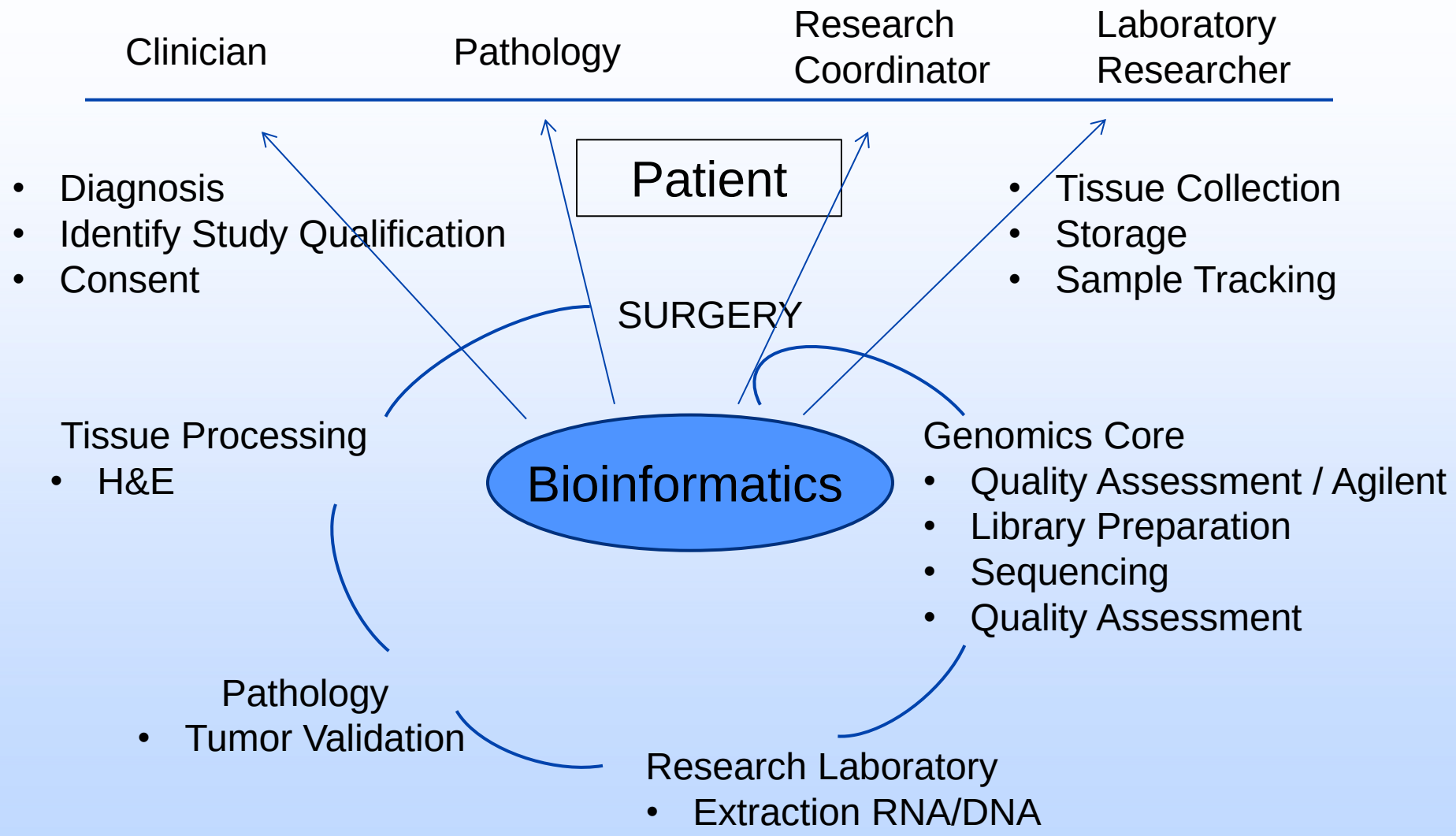
Head and Neck NGS Project Workflow



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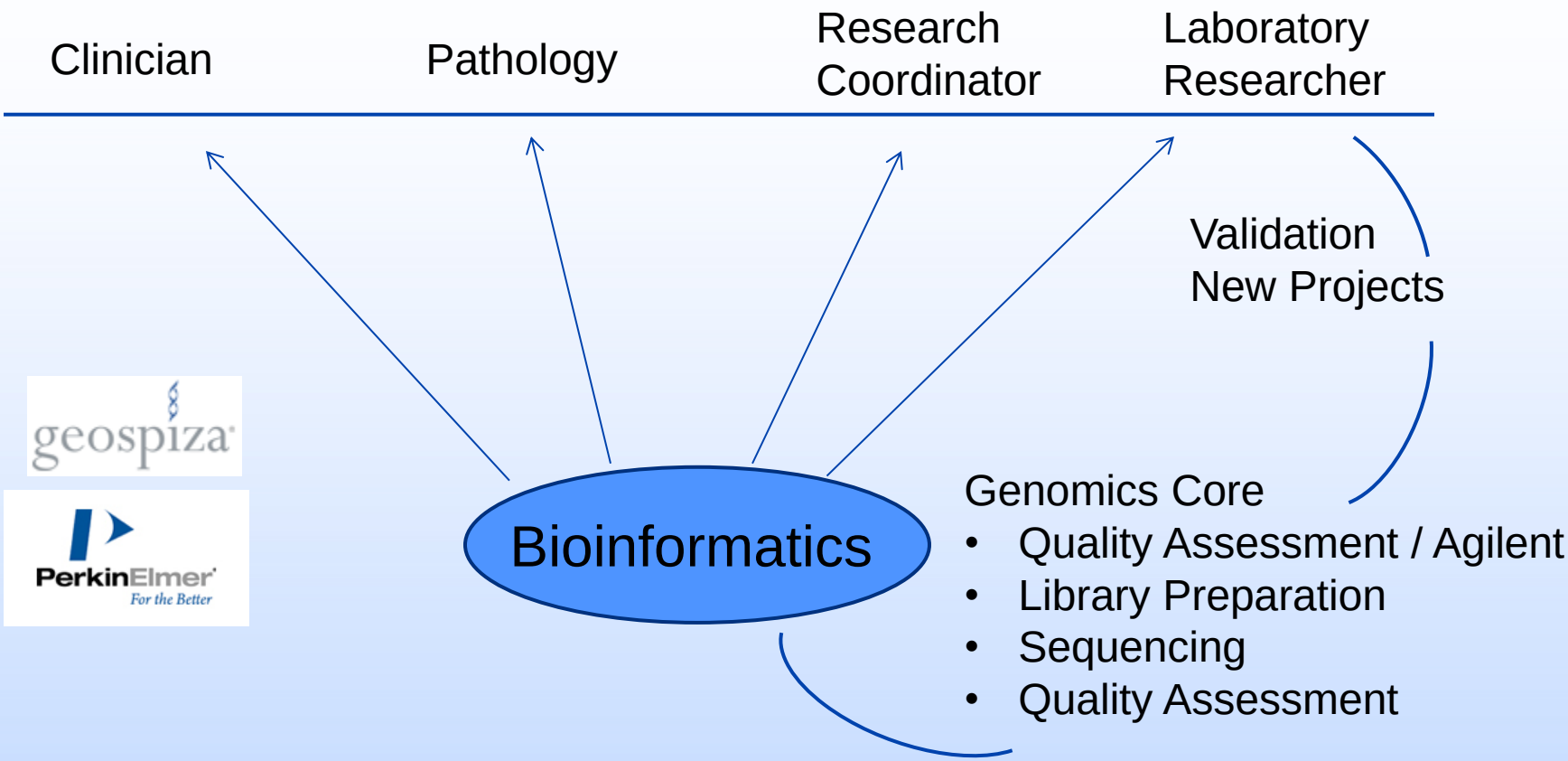
Head and Neck NGS Project Workflow



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Why GeneSifter?

- Secure, Cloud based
- Data Sharing between multiple users
- Easy access to data
  - Standard Computer, iPad, Smartphone
- Intuitive Interface
  - Does not require extensive experience with analysis
- Ability to visualize data
  - Mapped reads
  - Called variants
- Integrated analysis tools
  - Pathway analysis
- Platform / Application independent
  - Allows combined analysis of multiple datasets

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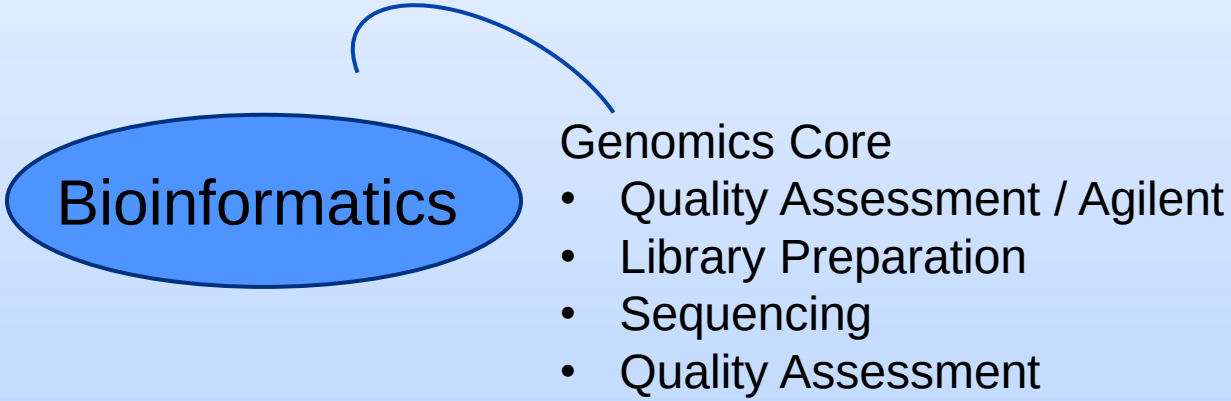
GeneSifter in the Genomics Core

- Upload the fastq reads generated
- Run alignment and quality pipelines
- Assess the post alignment sequence quality over entire read length
- Remove data from account



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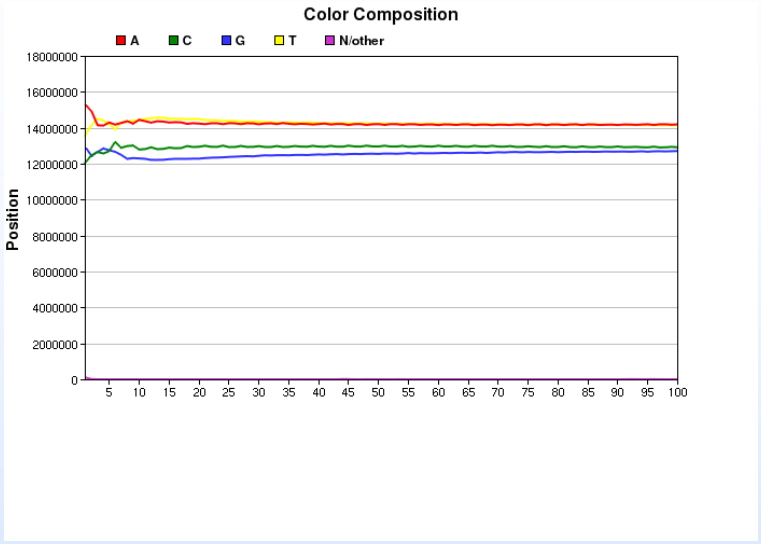
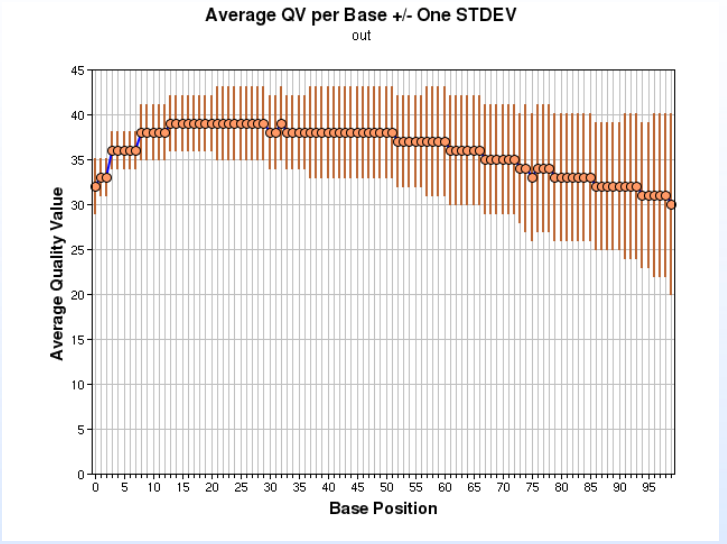
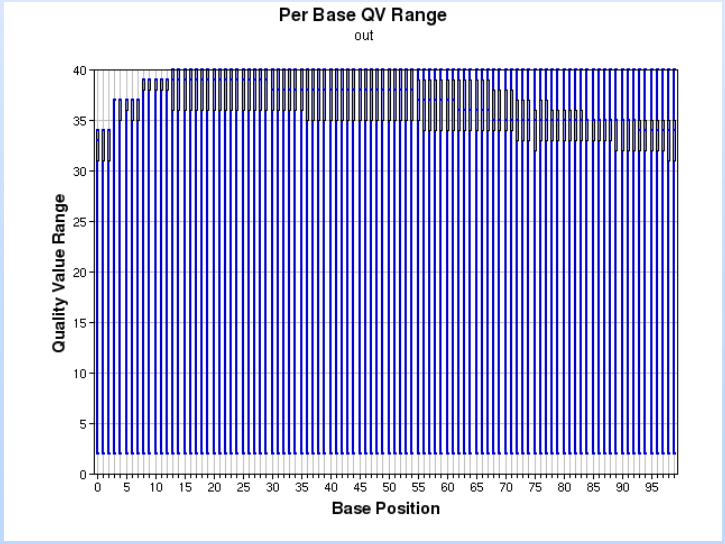
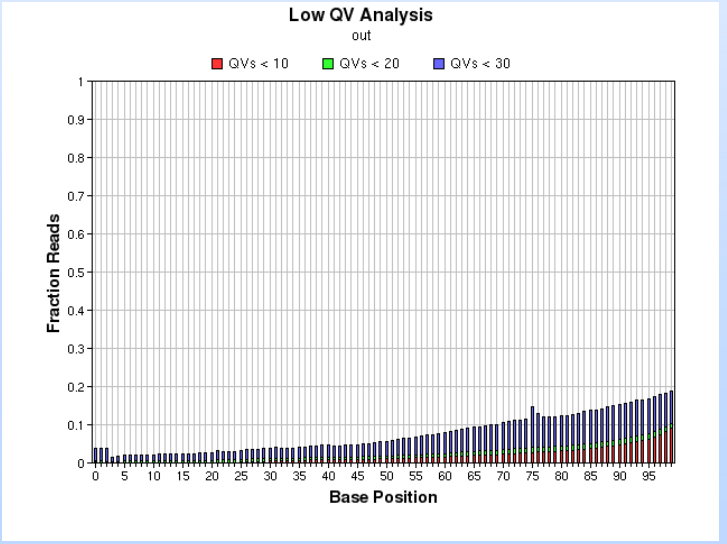
GeneSifter in the Genomics Core









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Clinical Aims

- Develop markers for patient stratification / risk factors
- Develop tools for early detection of primary/recurrent disease

Clinical Funding

- Clinicians
- Residents

Clinical

Head and Neck NGS Project

Basic / Bench

Peer Review Funding

- Faculty
- Postdocs / Students
- Technicians

Basic Research

- Mechanisms of transcript dysregulation
- RNA editing
- Identify fusion transcripts

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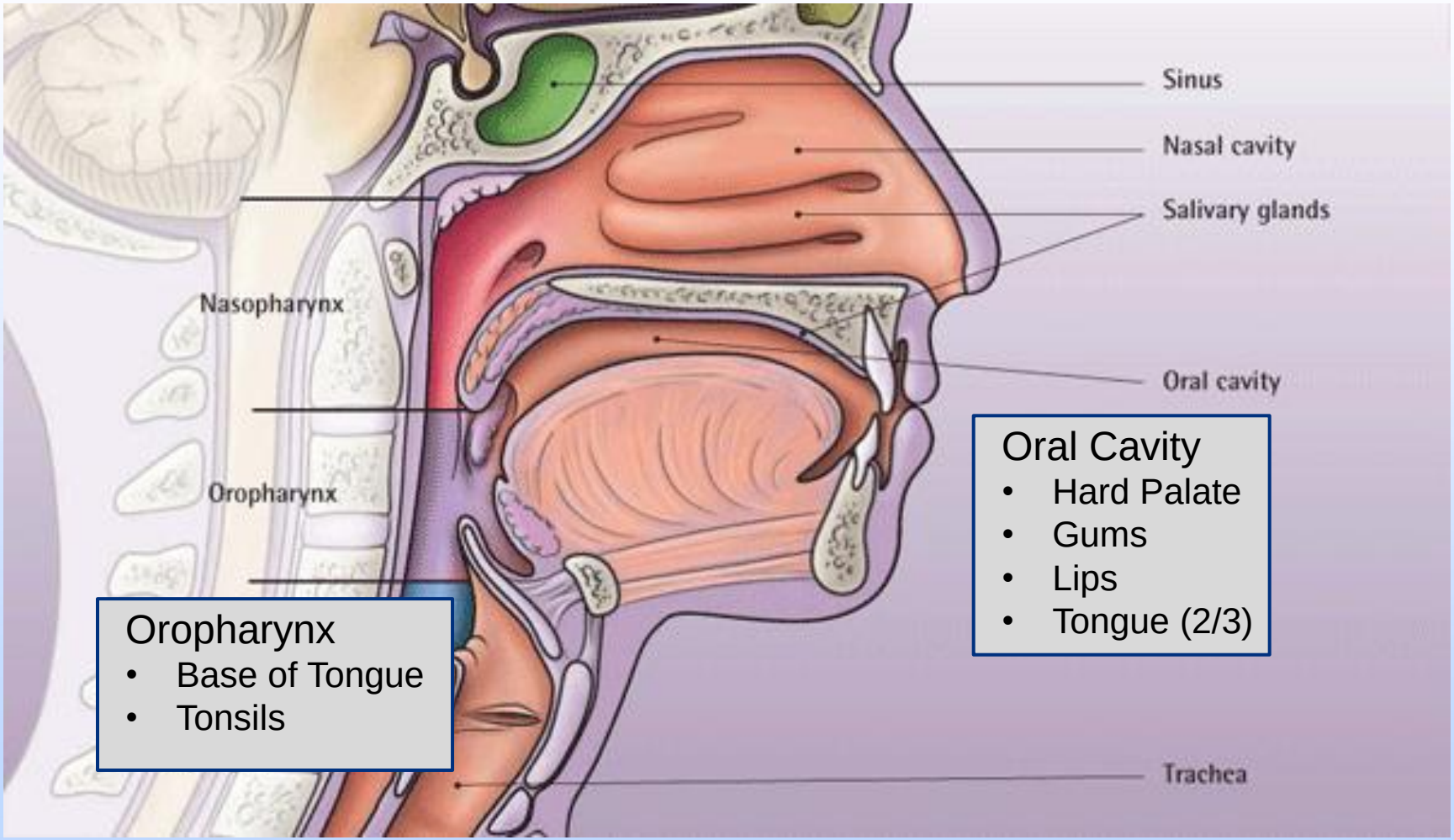
Head and Neck Cancer



Oral Cavity
- Hard Palate
- Gums
- Lips
- Tongue (2/3)

Oropharynx
- Base of Tongue
- Tonsils

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Head and Neck Cancer

- 6th most common cancer worldwide
- 95% squamous cell carcinoma (SCC)
- 50% rate of 5 year survival for advanced disease
- Diagnosis – lesion detection, lacking tools for early detection
- Treatment – Surgery, radiation, chemotherapy

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Risk Factors

- Smoking and Drinking
  - 6-7th decade of life, prolonged exposure

- HPV (16,18)
  - Oropharyngeal SCC (30-90% all patients)
  - Younger Patients (<50 years)
  - Lack traditional risk factors
  - Chemo/Radiation sensitive

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Patient Stratification

- HPV positive OPSCC
  - Experience better treatment response
  - Ideal candidates for robotic surgical approach
  - Interest in deescalated treatment in patients with HPV associated tumors

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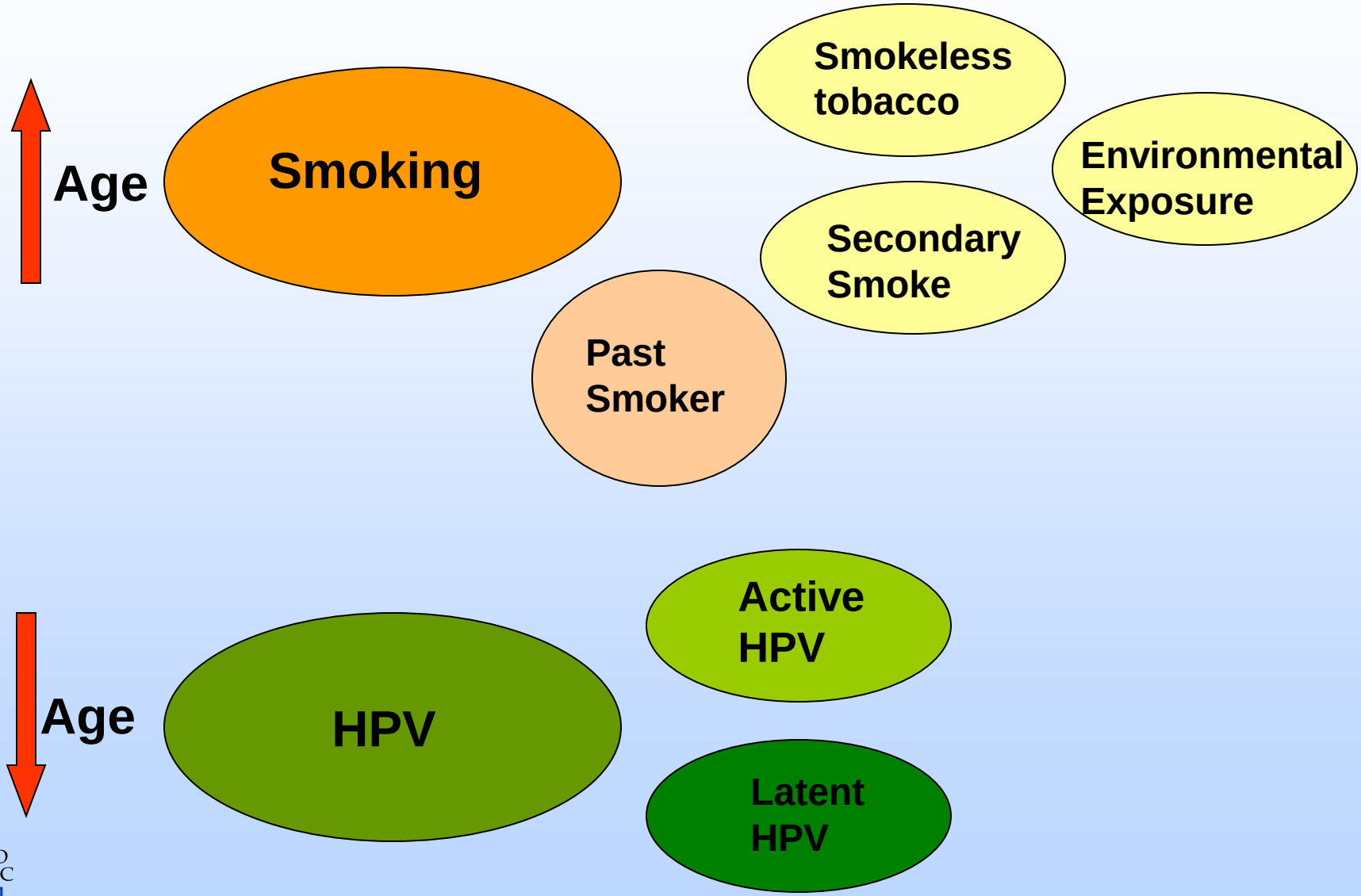
Patient Stratification



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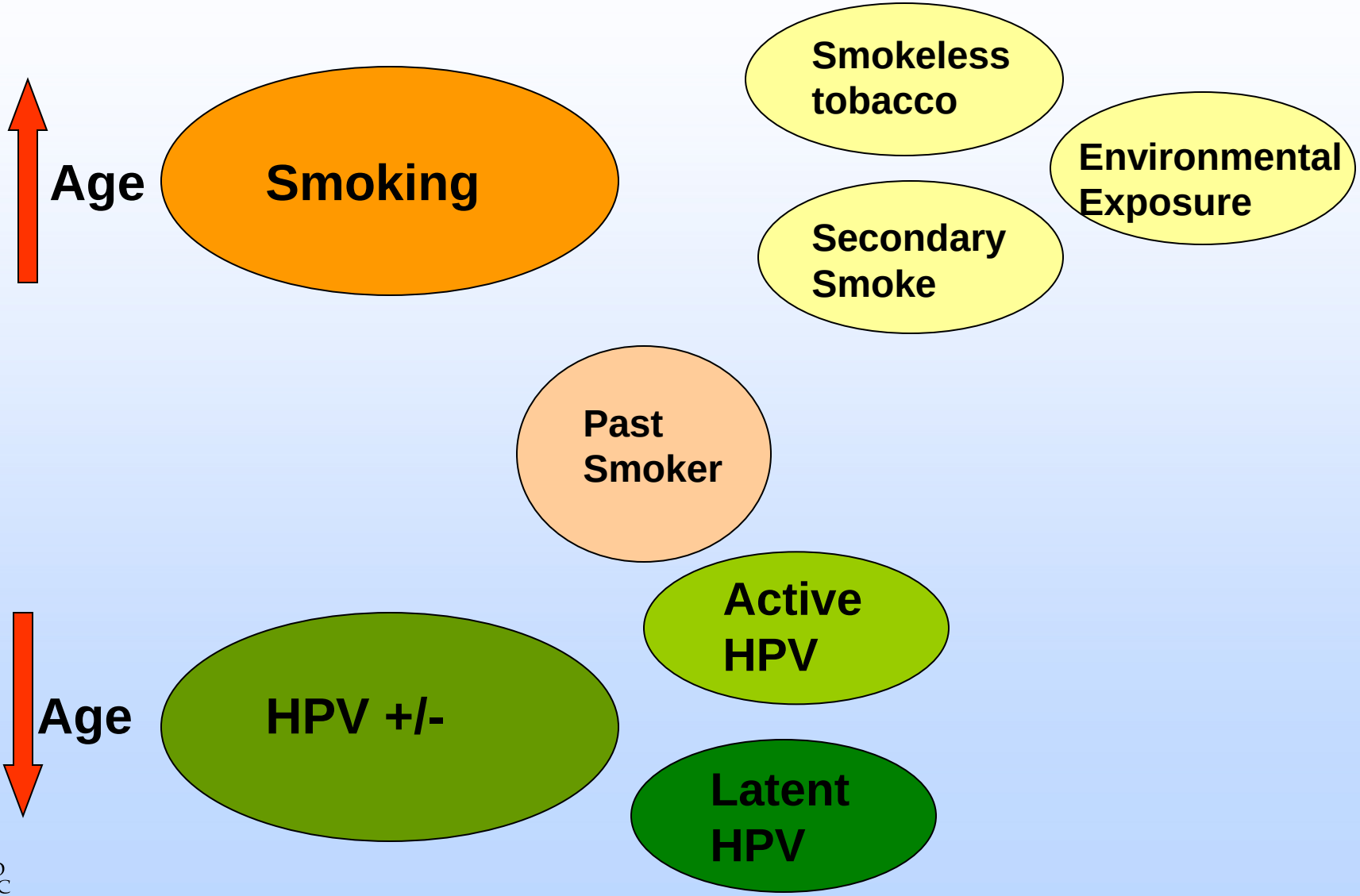
Which factors are most significant for the development of disease?



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Why choose NGS

- Preliminary data – microarray, aCGH
- NGS
  - Gene expression
  - Variant detection
  - Fusion transcripts
  - RNA editing
- mRNA-Seq
- Exome enrichment, NGS

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mRNA-Seq transcriptional profiling

- 18 Tumor / Patient Match Normal Tissues
- Illumina GAIIx
  - ~65 million reads per sample
- Data analysis
  - GeneSifter, Geospiza, PerkinElmer

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Patient Demographics

TABLE 2. Patient Demographics Grouped by Smoking Status

Patient No.	Age (y)	Sex	Subsite	HPV-16 test result	T stage	N stage	M stage	Grade
Current smokers								
1	56	M	Tonsil	Positive	1	0	0	4
2	73	M	Tonsil	Negative	4a	2c	0	3
3	54	M	Base of tongue	Negative	4	2b	0	3
Former smokers								
4	48	M	Tonsil	Positive	4a	2c	0	3
5	64	F	Tonsil	Positive	1	2a	0	4
6	66	M	Base of tongue	Positive	2	2b	0	3
Never smokers								
7	46	M	Tonsil	Positive	3	2b	1	3
8	49	F	Tonsil	Positive	2	0	0	4
9	73	M	Tonsil	Negative	2	2b	0	3

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Laborde RR, Wang VW, Smith TM, Olson NE, Olsen SM, García JJ, Olsen KD, Moore EJ, Kasperbauer JL, Tombers NM, Smith DI. 2012. Transcriptional profiling by sequencing of oropharyngeal cancer. *Mayo Clin Proc*. Mar;87(3):226-32.

mRNA-Seq data in Excel

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S
1	Index	Chromosor	Start	End	Genes	Smith10.C	Smith10.C	Smith11.C	Smith11.C	Smith12.C	Smith12.C	Smith13.C	Smith13.C	Smith14.C	Smith14.C	Smith15.C	Smith15.C	Smith16.C	Smith16.C	Sn
2																				
3	0	1	815	19919	LOC65363	0.49503	2986	0.4317	2604	0.48873	2948	0.28084	1694	0.40849	2464	0.01127	68	0.63064	3804	(
4																				
5																				
6	12	1	663007										11	0.00783	25	0.00094	3	0.01097	35	(
7																				
8	13	1	742614										4	0.00362	3	0	0	0.00845	7	(
9																				
10	14	1	751449										23	0.03037	39	0.00467	6	0.03583	46	(
11																				
12	15	1	845464										8	0.037	33	0	0	0.01009	9	
13																				
14	16	1	850984										135	0.05885	128	0	0	0.01471	32	(
15																				
16	17	1	869825										3849	1.55266	3759	0.02602	63	1.52127	3683	1
17																				
18	18	1	885830	890958	KLHL17	0.28633	733	0.32188	824	0.35508	909	0.19258	493	0.2332	597	0.00586	15	0.59883	1533	(
19																				
20	19	1	891740	900351	PLEKHN1	1.23918	2979	0.26498	637	0.18386	442	0.58444	1405	0.21298	512	0.00125	3	1.62521	3907	
21																				
22	20	1	900442	907330	C1orf170	0.559	1696	0.1002	304	0.86618	2628	0.19281	585	1.82531	5538	0	0	0.54219	1645	(
23																				
24	21	1	924207	9									815	0.64351	565	0.00683	6	0.67882	596	1
25																				
26	22	1	938742										2063	0.79495	504	0.04416	28	1.45584	923	1
27																				
28	23	1	945366										4171	0.99453	7279	0.01626	119	1.06599	7802	2
29																				
30	24	1	1007068										337	0.11419	367	0.00342	11	0.22091	710	(
31																				
32	25	1	1100820										3	0.00881	9	0	0	0.00196	2	(
33																				
34	26	1	1104940										13	0.009	19	0.00095	2	0.02511	53	(
35																				
36	27	1	1128751	1131952	TNFRSF1	0.2125	255	4.01667	4820	0.62417	749	0.97083	1165	0.31667	380	0.01167	14	0.205	246	2
37																				
38	28	1	1136569	1139375	TNFRSF4	0.06929	74	0.12547	134	0.41292	441	0.06929	74	0.12547	134	0	0	0.04401	47	(
39																				
40	29	1	1142151	1157274	SDF4	2.67364	5620	2.6118	5490	2.26832	4768	2.98858	6282	2.47431	5201	0.06232	131	1.79924	3782	2
41																				
42	30	1	1157492	1160284	B3GALT6	0.30326	847	0.39957	1116	0.32975	921	0.54386	1519	0.40351	1127	0.00609	17	0.29896	835	(
43																				
44	31	1	1167696	1171965	FAM132A	0.05405	56	2.17954	2258	0.04923	51	0.06467	67	0.04633	48	0	0	0.05985	62	(
45																				

- 51255 Rows
- Export for ANOVA, T-tests
- Large files, difficult to share with collaborators

- Generated Lists of specific targets
- Repeated analysis for new comparisons
- Fewer options for data visualization

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GeneSifter

Start an Evaluation
>>NGS Analysis
>>Microarray Analysis

Watch Webinars

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Product Updates

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877.WEB.GENE

Upcoming Events

Webinar: Novel Approaches to Automated Sample Prep and Data Analysis for RNA-Seq
May 30, 2012 1pm EST, 10am PST

PerkinElmer Owners Group Meeting (USA - East Coast)
June 7-8, 2012 Newton, MA

PerkinElmer Owners Group Meeting (Europe)
June 12-13, 2012 London, UK

Geospiza develops software systems to accelerate genetic research. Built by biologists for biologists, our products help you quickly see the science.

Data Analysis
Easily visualize and mine the mountain of genetic data produced by Microarray and Next Generation Sequencing (NGS) technology.

LIMS System
Get complex laboratory workflows, data management and advance commercial billing for less time and cost of other LIMS systems.

DNA Sequencing Services
Speed up your research and go from samples to results in as little as 4-6 wks. On-Demand sequencing, data analysis and data storage meld into one complete unit.

FINCH Talk!
Industry blog by founder and CSO, Todd Smith

Presentations
Watch a quick video about of our LIMS and Data Analysis solutions

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Upload data

QuickLoad Wizard

Description:
Use this tool to load **Affymetrix** (CHP or tab-delimited text files), **Illumina** or **CodeLink** arrays.

Run QuickLoad Wizard

Batch Upload

Description:
The Batch Upload wizard accepts a spreadsheet, in tab-delimited text format, containing data from several different experiments.

Run Batch Upload

FlexLoad Wizard

Description:
Using the FlexLoad Wizard will allow you to create and save custom templates for loading almost any other type of file. Users of **Agilent**, **2-color**, or **custom chips** should use this upload tool.

Run FlexLoad Wizard

Advanced Upload Methods

Description:
This **Affymetrix**-specific tool allows you to load **CEL files** and perform **RMA**, **GC-RMA** or **MAS5** on the probe-level data.

Run Advanced Upload Methods

Next Gen File Upload

Description:
Use this tool to load **Next Gen** sequencing data files for analysis.

Next Gen File Upload

Next Gen File Upload

Step 1 of 3

Type the name of a new folder or select an existing folder for your data. This field is not required.

Directory:

--OR-- <Select target folder>

File Type:

Auto-Detect Characterize files after upload is finished?

Cancel Next

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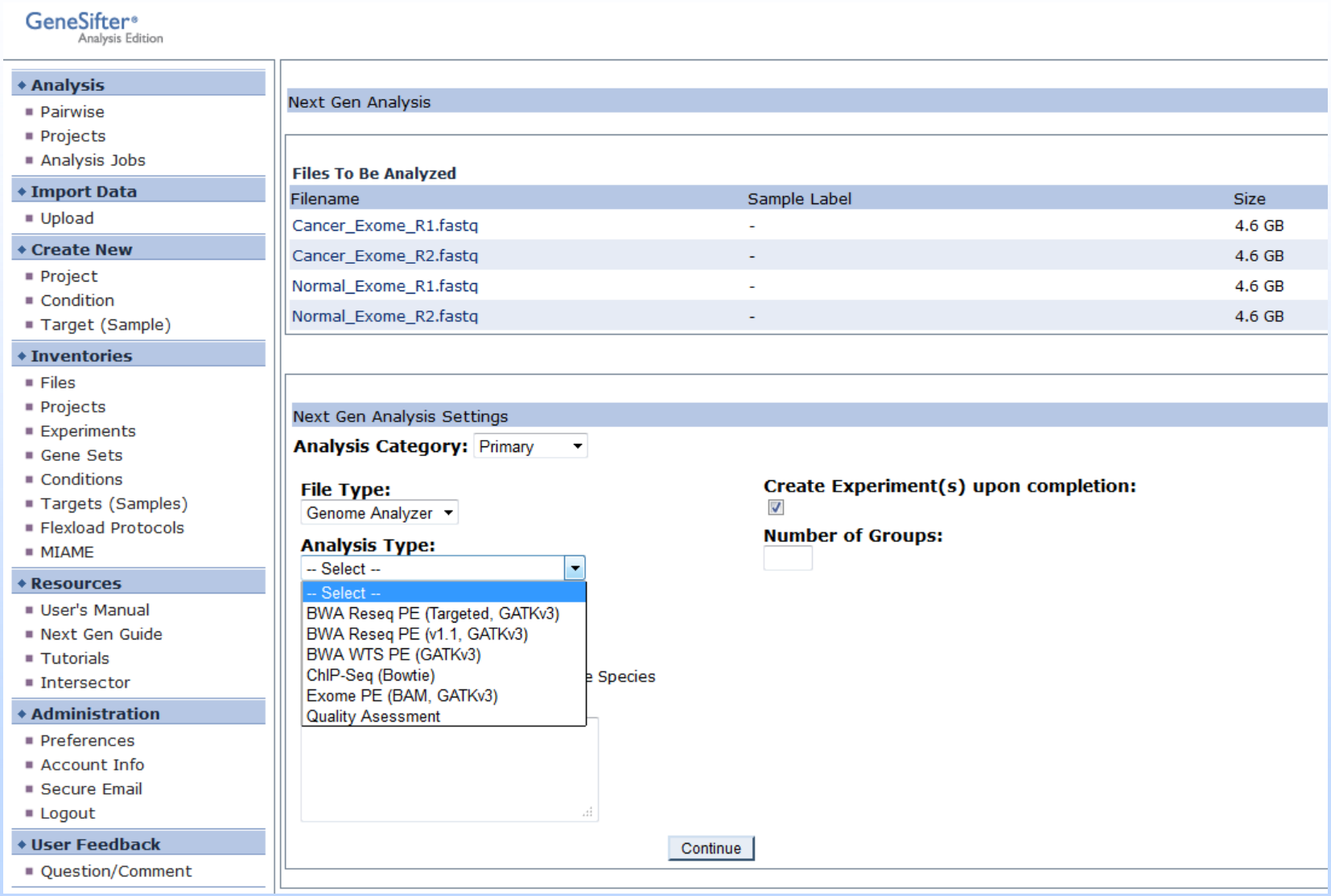
Initiate Analysis Pipeline



Create Project

- Analysis
  - Pairwise
  - Projects
  - Analysis Jobs
- Import Data
  - Upload
- Create New
  - Project
  - Condition
  - Target (Sample)
- Inventories
  - Files
  - Projects
  - Experiments
  - Gene Sets
  - Conditions
  - Targets (Samples)
  - Flexload Protocols
  - MIAME
- Resources
  - User's Manual
  - Next Gen Guide
  - Tutorials
  - Intersector
- Administration
  - Preferences
  - Account Info
  - Secure Email
  - Logout
- User Feedback
  - Question/Comment

» Create New Project - Choose Method

Project analysis set up organizes array data for making 3 or more simultaneous comparisons.

Create project using conditions

Set up a multi comparison analysis by assigning individual gene sets/samples to groups using already defined conditions. Typically used for comparing multiple treatments and timecourses with or without replicates for each condition.

Create project using conditions...

Create project using targets

Assign individual gene sets/samples to groups using already defined targets for a multi comparison analysis. Typically used for comparing multiple patients, animals, etc. with or without replicates for each target.

Create projects using targets...

Create project using new categories

This is a flexible option to assign gene sets/samples to novel groups. Set up a multi comparison analysis by assigning arrays/samples to groups irrespective of defined condition or target.

Create project using new categories...

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◆ Analysis
- Pairwise
- Projects
- Analysis Jobs

◆ Import Data
- Upload

◆ Create New
- Project
- Condition
- Target (Sample)

◆ Inventories
- Files
- Projects
- Experiments
- Gene Sets
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- MIAME

◆ Resources
- User's Manual
- Next Gen Guide
- Tutorials
- Intersector

◆ Administration
- Preferences
- Account Info
- Secure Email
- Logout

◆ User Feedback
- Question/Comment

Main (login: mayo_project) > Create New > Project

» Create New Project (Step 1 of 3)
Select Gene Set

Project Title: ANOVA_Oroph_Risk

Description: Oropharyngeal Paired Samples

Gene Set: Human Whole Transcriptome: Human Whole Transcr

Number of categories: 6

Continue

GeneSifter® Analysis Edition

Home | Support | Geospiza | Logout

- **Analysis**
  - Pairwise
  - Projects
  - Analysis Jobs
- **Import Data**
  - Upload
- **Create New**
  - Project
  - Condition
  - Target (Sample)
- **Inventories**
  - Files
  - Projects
  - Experiments
  - Gene Sets
  - Conditions
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  - User's Manual
  - Next Gen Guide
  - Tutorials
  - Intersector
- **Administration**
  - Preferences
  - Account Info
  - Secure Email
  - Logout
- **User Feedback**
  - Question/Comment

Main (login: mayo_project) > Create New > Project

» Create New Project (Step 2 of 3): Select normalization, data transformation and create categories

Title: ANOVA_Oroph_Risk

Gene Set: Human Whole Transcriptome

Normalization: Mapped Reads (NGS)

MA = Microarray
NGS = Next Gen

Data Transformation: No transformation

Category 1: Current Smoking Tumor

Category 2: Current Smoking Normal

Category 3: Former Smoking Tumor

Category 4: Former Smoking Normal

Category 5: Never Smoking Tumor

Category 6: Never Smoking Normal

Continue

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» Create New Project (Step 3 of 3)
Select Experiments To Include For Each Category

Title: ANOVA_Oroph_Risk
Normalization: mapped_reads
Data Transformation: None

Category	Experiment	Target	Condition
Never Smoking Normal	Smith10	10 N YN	Normal
Current Smoking Normal	Smith12	12 N OS	Normal
None	Smith14	14 N YP	Normal
Former Smoking Normal	Smith18	18 N OP	Normal
Never Smoking Normal	Smith2	2 N YN	Normal
Former Smoking Normal	Smith20	20 N OP	Normal
Never Smoking Normal	Smith22	22 N ON	Normal
Current Smoking Normal	Smith24	24 N OP	Normal
Former Smoking Normal	Smith4	4 N YP	Normal
Current Smoking Normal	Smith6	6 N OS	Normal
None	Smith8	8 N OS	Normal
Never Smoking Tumor	Smith1	1 T YN	Tumor
Current Smoking Tumor	Smith11	11 T OS	Tumor
None	Smith13	13 T YP	Tumor
Former Smoking Tumor	Smith17	17 T OP	Tumor
Former Smoking Tumor	Smith19	19 T OP	Tumor
Never Smoking Tumor	Smith21	21 T ON	Tumor
None	Smith23	23 T OP	Tumor
Former Smoking Tumor	Smith3	3 T YP	Tumor
Current Smoking Tumor	Smith5	5 T OS	Tumor
Current Smoking Tumor	Smith7	7 T OS	Tumor
Never Smoking Tumor	Smith9	9 T YN	Tumor

Continue

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- Current Smoking Tumor [Control]
- Current Smoking Normal
- Former Smoking Tumor
- Former
- Never
- Never

GeneSifter® Analysis Edition

Show All Ger

Project: ANOVA_Oroph_Risk

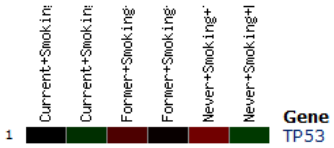
(login: mayo_project)> Projects > Analysis > Gene Navigation > Results

Identifiers: TP53 | No Statistics

Show: 20 | Quality: N/A | Search | (1 result found) [1 - 1]

[**Reports:** Ontology | KEGG | Chromosome] [**Cluster:** Samples (r) | Genes (r) | Samples + Genes (r) | PCA | PCA2] [**Results:** Export | Save | View As Image]

Legend: 0.25 — 1 — 4



Sort: By Expression | Statistics: No Statistics | Show: 20 | Search | Save

Search by Gene Symbol

Identifiers:

TP53

Sort: By Expression | Statistics: No Statistics | Show: 20 | Search | Save

Search by Chromosome

Chromosome: *

Sort: By Expression | Statistics: No Statistics | Show: 20 | Search

» Gene Summary: Tumor protein p53

» One-Click Gene Summary™

Entrez Gene: 7157
Cluster ID: Hs.654481
UG Title: Tumor protein p53
Gene ID: TP53
Chromosome: 17
Cytoband: 17p13.1
Seq Count: 786
Entrez Gene: 7157
Gene Name: tumor protein p53
Synonyms: FLJ92943 | LFS1 | P53 | TRP53
Summary: This gene encodes tumor protein p53, which responds to diverse cellular stresses to regulate target genes that induce cell cycle arrest, apoptosis, senescence, DNA repair, or changes in metabolism. p53 protein is expressed at low level in normal cells and at a high level in a variety of transformed cell lines, where it's believed to contribute to transformation and malignancy. p53 is a DNA-binding protein containing transcription activation, DNA-binding, and oligomerization domains. It is postulated to bind to a p53-binding site and activate expression of downstream genes that inhibit growth and/or invasion, and thus function as a tumor suppressor. Mutants of p53 that frequently occur in a number of different human cancers fail to bind the consensus DNA binding site, and hence cause the loss of tumor suppressor activity. Alterations of this gene occur not only as somatic mutations in human malignancies, but also as germline mutations in some cancer-prone families with Li-Fraumeni syndrome. Multiple p53 variants due to alternative promoters and multiple alternative splicing have been found. These variants encode distinct isoforms, which can regulate p53 transcriptional activity. [provided by RefSeq]

TP53 Search PubMed

Search for Homologs:

Select a project Search

OMIM:

- NASOPHARYNGEAL CARCINOMA
- PANCREATIC CANCER
- LI-FRAUMENI SYNDROME 1; LFS1
- BREAST CANCER
- HEPATOCELLULAR CARCINOMA
- OSTEOGENIC SARCOMA
- PAPILLOMA OF CHOROID PLEXUS
- NASOPHARYNGEAL CARCINOMA, SUSCEPTIBILITY TO, 2
- ADRENOCORTICAL CARCINOMA, HEREDITARY; ADCC
- COLORECTAL CANCER; CRC

Gene Ontologies:

Biological Process
- activation of caspase activity by cytochrome c
- apoptosis
- base-excision repair
- blood coagulation
- cell aging
- cell cycle
- cell cycle arrest
- cell cycle checkpoint
- cell differentiation
- cell proliferation
- cellular protein localization
- cellular response to glucose starvation
- cellular response to ionizing radiation
- cellular response to UV
- determination of adult lifespan
- DNA damage response, signal transduction by p53 class mediator
- DNA damage response, signal transduction by p53 class mediator resulting in cell cycle arrest
- DNA damage response, signal transduction by p53 class mediator resulting in induction of apoptosis
- DNA damage response, signal transduction by p53 class mediator resulting in transcription of p21 cla
- DNA strand renaturation
- ER overload response
- induction of apoptosis by intracellular signals
- interspecies interaction between organisms
- mitotic cell cycle G1/S transition DNA damage checkpoint
- multicellular organismal development
- negative regulation of apoptosis
- negative regulation of cell growth
- negative regulation of cell proliferation
- negative regulation of gene-specific transcription from RNA polymerase II promoter
- negative regulation of helicase activity
- negative regulation of transcription from RNA polymerase II promoter
- nucleotide-excision repair
- oxidative stress-induced premature senescence
- positive regulation of gene-specific transcription from RNA polymerase II promoter
- positive regulation of histone deacetylation
- positive regulation of neuron apoptosis
- positive regulation of peptidyl-tyrosine phosphorylation
- positive regulation of thymocyte apoptosis
- positive regulation of transcription from RNA polymerase II promoter
- protein complex assembly
- protein localization
- protein tetramerization
- Ras protein signal transduction
- regulation of apoptosis

- Current Smoking Tumor [Control]
- Current Smoking Normal
- Former Smoking Tumor
- Former Smoking Normal
- Never Smoking Tumor
- Never Smoking Normal

Show All Genes

(login: mayo_project) > Analysis > Projects

Project Analysis: ANOVA_Oroph_Risk

[**Gene Navigation** | Gene Function | Pattern Navigation | Cluster | Export | Two-Way Anova]

Search by Name

Name:

Match: All Words Sort: By Expression Statistics: No Statistics Show: 20 Search

Search by Accession, UniGene, Entrez, Affy Probe Set, Illumina or CodeLink ID

Identifiers:

Saved Searches: Select

Affy ST IDs

Sort: By Expression Statistics: No Statistics Show: 20 Search Save

Search by Gene Symbol

Identifiers:

Sort: By Expression Statistics: No Statistics Show: 20 Search Save

Search by Chromosome

Chromosome: *

Sort: By Expression Statistics: No Statistics Show: 20 Search

Project: ANOVA_Oroph_Risk

(login: mayo_project) > Projects > Analysis > Pattern Navigation > Results

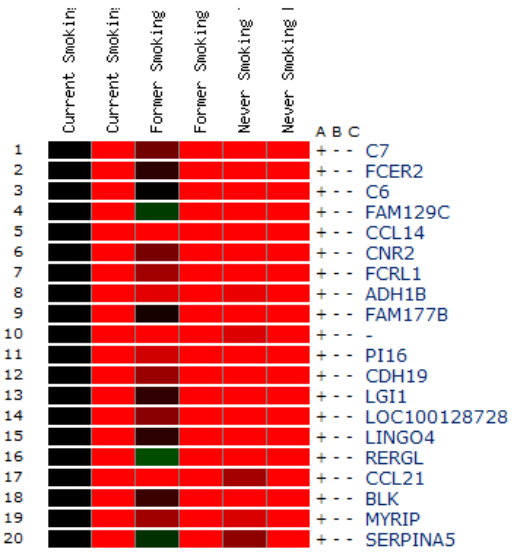
Display Genes That Pass: Any

Show: 20 p Cutoff: 0.05 Search (5341 results found) [1 - 20] [21 - 40]

[**Reports:** Ontology | KEGG | Chromosome] [**Cluster:** Samples (r) | Genes (r) | Samples + Genes (r) | PCA | PCA 3d] [**Results:** Export | Save | View As Image

A: Disease
B: Risk Factor
C: Interaction

Legend: 0.25 1



Display Genes That Pass: Any

Show: 20 p Cutoff: 0.05 Search (5341 results found) [1 - 20] [21 - 40]

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Transcriptional Profiling

- Differentially regulated genes
  - HOX, MMPs, T cell signaling, Immune responsive gene targets
- Regulatory Non-coding Transcripts
- Pathway analysis (GeneSifter)
  - Is there a difference between groups divided by risk factor?

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Condition 1 : Current Smoking Tumor
Condition 2 : Current Smoking Normal
Condition 3 : Former Smoking Tumor
Condition 4 : Former Smoking Normal
Condition 5 : Never Smoking Tumor
Condition 6 : Never Smoking Normal

Export Report

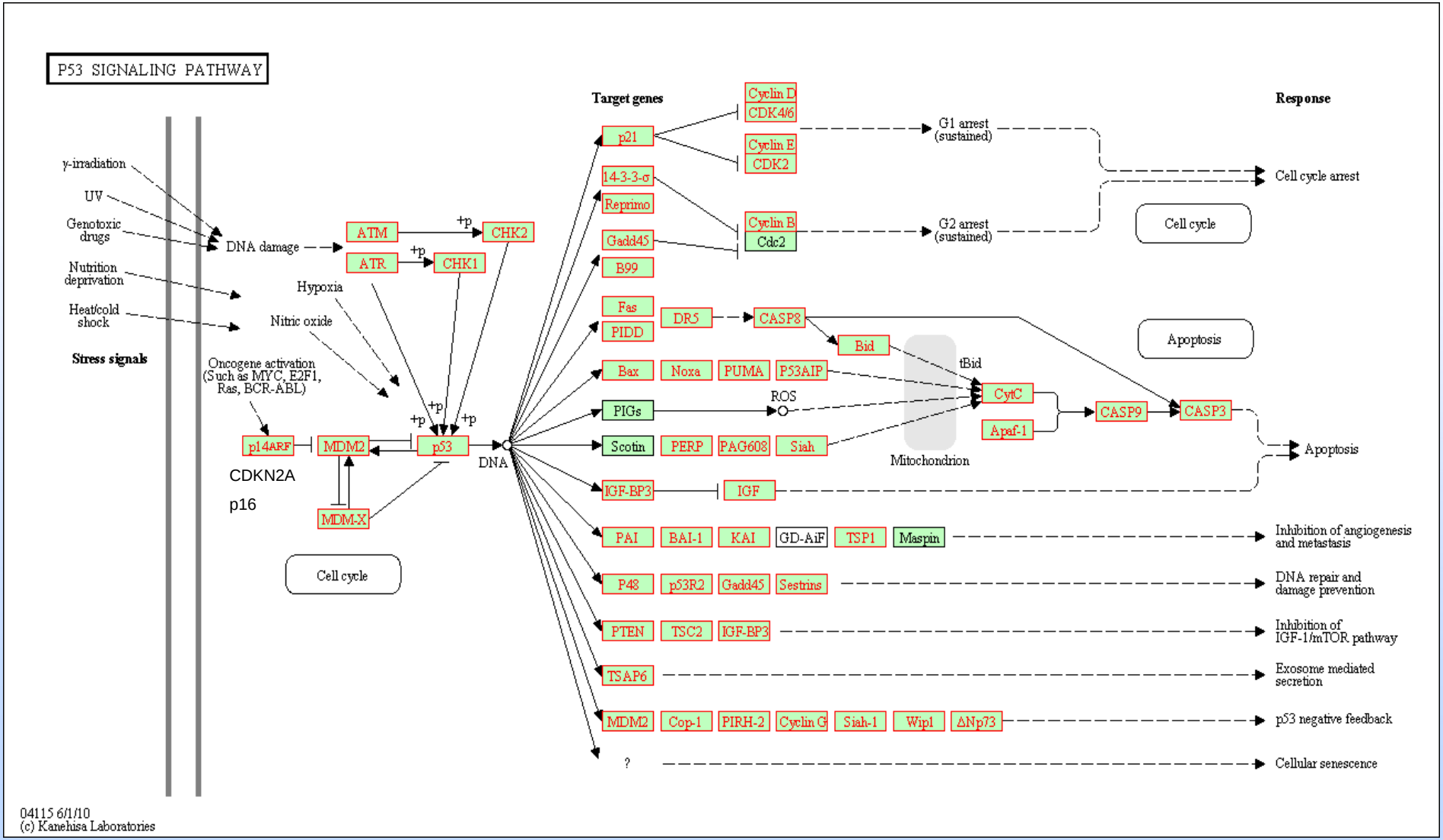
Project: ANOVA_C

A: D
B: R
C: I

Pathway	Genes	KEGG	Totals List	Totals Gene Set	z-score
Pathways in cancer			96	326	2.02
Cell cycle			76	124	9.53
RNA transport			75	156	6.84
Neuroactive ligand-receptor interaction			57	317	-2.87
Purine metabolism			55	161	2.81
Spliceosome			53	126	4.55
Cytokine-cytokine receptor interaction			51	275	-2.44
MAPK signaling pathway			51	271	-2.32
Systemic lupus erythematosus			50	135	3.35
Ubiquitin mediated proteolysis			48	135	2.94
Oocyte meiosis			46	112	4.04
Pyrimidine metabolism			43	99	4.35
Small cell lung cancer			36	85	3.79
Calcium signaling pathway			29	177	-2.62
DNA replication			28	36	7.40
p53 signaling pathway			28	68	3.16
Pancreatic cancer			25	70	2.14
Base excision repair			23	34	5.81
Proteasome			23	44	4.25
Nucleotide excision repair			22	44	3.90
Osteoclast differentiation			22	128	-2.00
Natural killer cell mediated cytotoxicity			20	134	-2.67
Tight junction			20	132	-2.58
Vascular smooth muscle contraction			18	125	-2.71
Aminoacyl-tRNA biosynthesis			17	41	2.49
Olfactory transduction			17	386	-9.58
Bladder cancer			16	42	2.01
Homologous recombination			16	28	3.98
Mismatch repair			16	23	4.99
Basal transcription factors			15	37	2.23
RNA polymerase			15	29	3.37
Pancreatic secretion			14	102	-2.60
Toll-like receptor signaling pathway			13	102	-2.83
Selenoamino acid metabolism			12	26	2.54
B cell receptor signaling pathway			11	75	-2.04
Drug metabolism - cytochrome P450			10	73	-2.20
One carbon pool by folate			10	18	3.03
RIG-I-like receptor signaling pathway			10	71	-2.09
Ribosome			8	89	-3.47
Glycerolipid metabolism			6	50	-2.10
Intestinal immune network for IgA production			5	46	-2.19
Fat digestion and absorption			4	45	-2.47
Viral myocarditis			4	68	-3.63
Allograft rejection			2	35	-2.62
Autoimmune thyroid disease			2	50	-3.41
Linoleic acid metabolism			2	28	-2.16
Type I diabetes mellitus			2	41	-2.96
Ascorbate and aldarate metabolism			1	26	-2.47
Graft-versus-host disease			1	37	-3.12

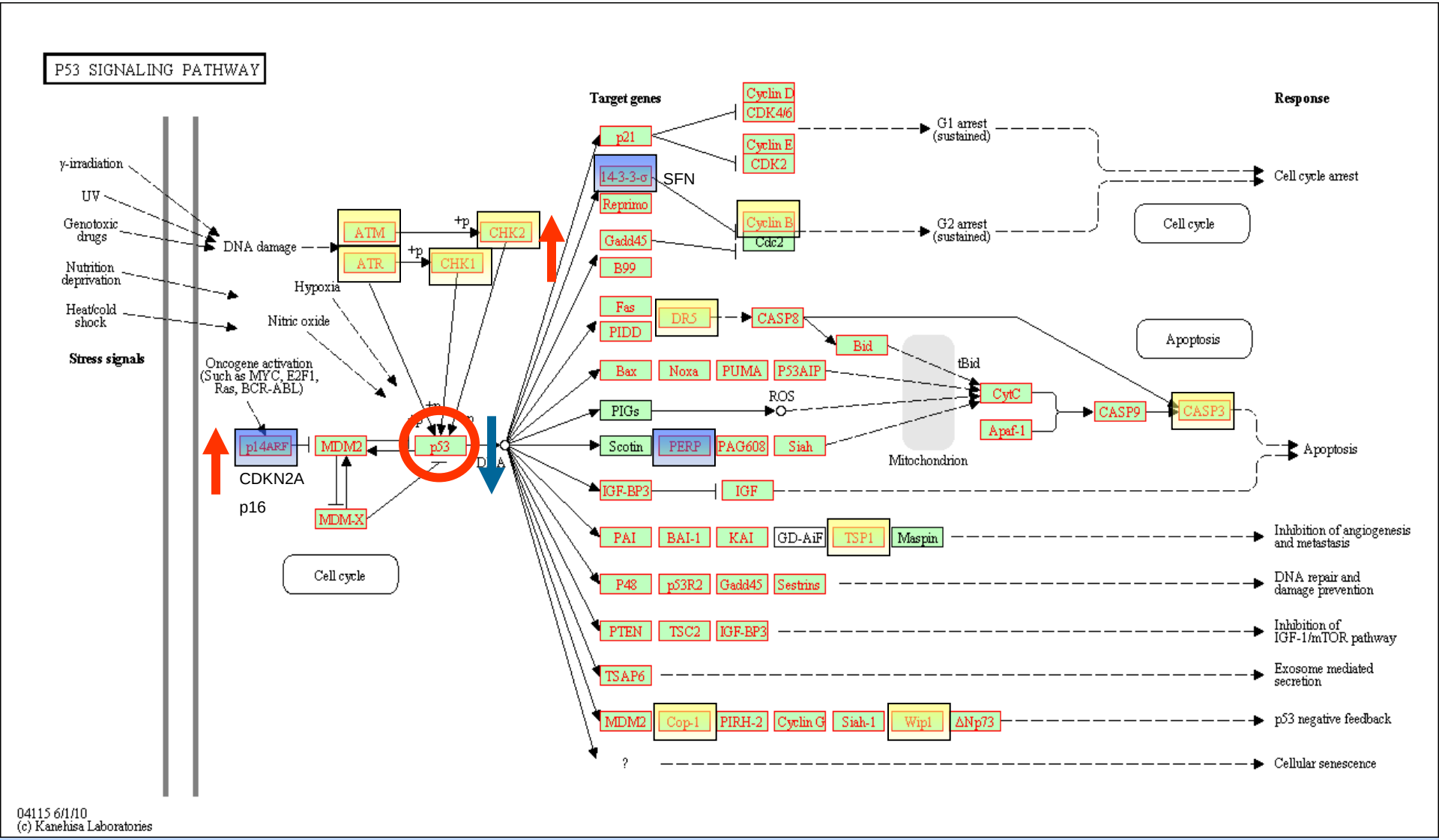
Pathway Analysis

Smoking Status



Current Smokers

Never smokers



Validation Assays

- Validate in larger number of tumor/normal pairs
- Nanostring nCounter digital counter
  - 96 gene codesets
  - 44 sample pairs
  - 100ng input RNA
- qPCR, Fluidigm also options

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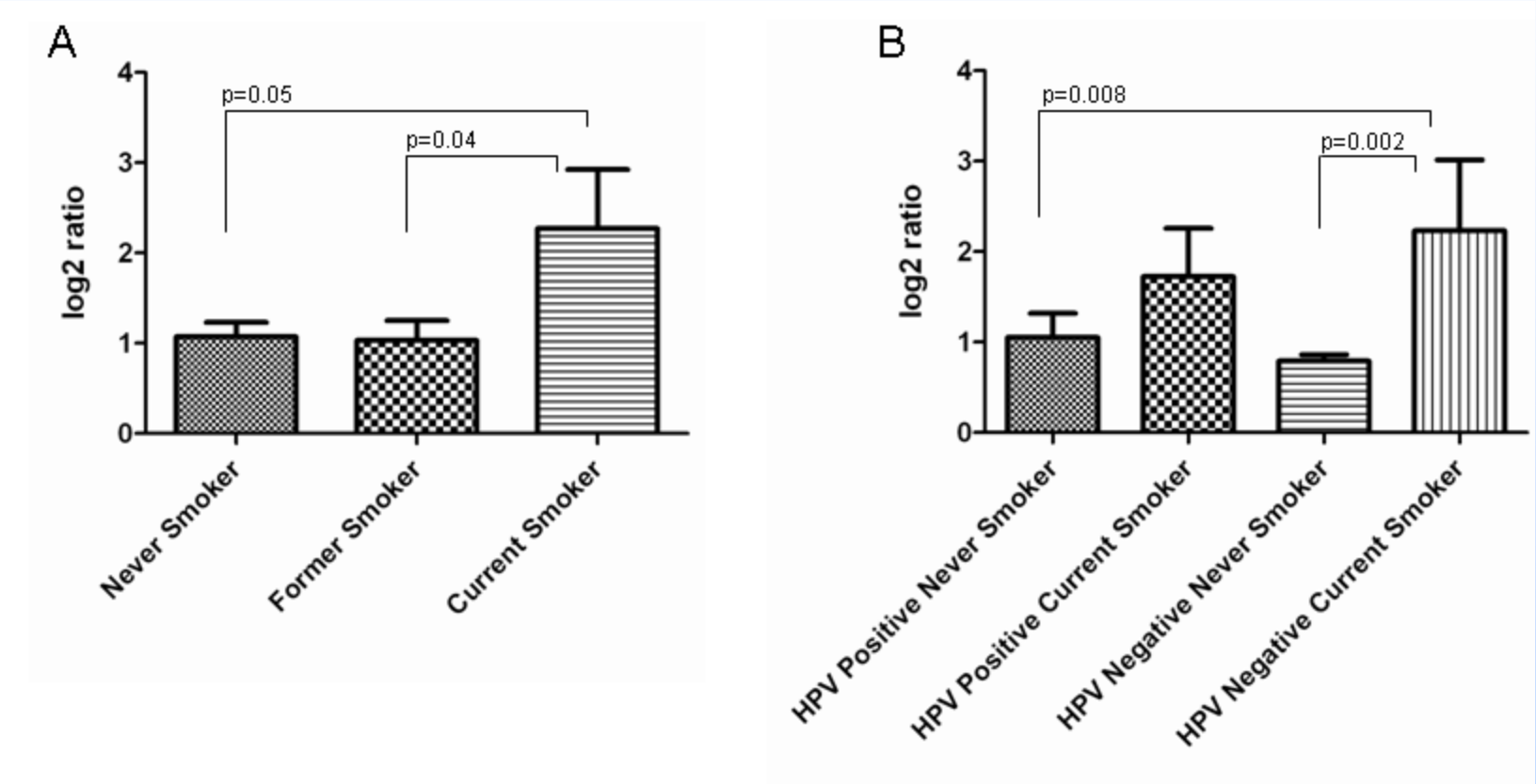
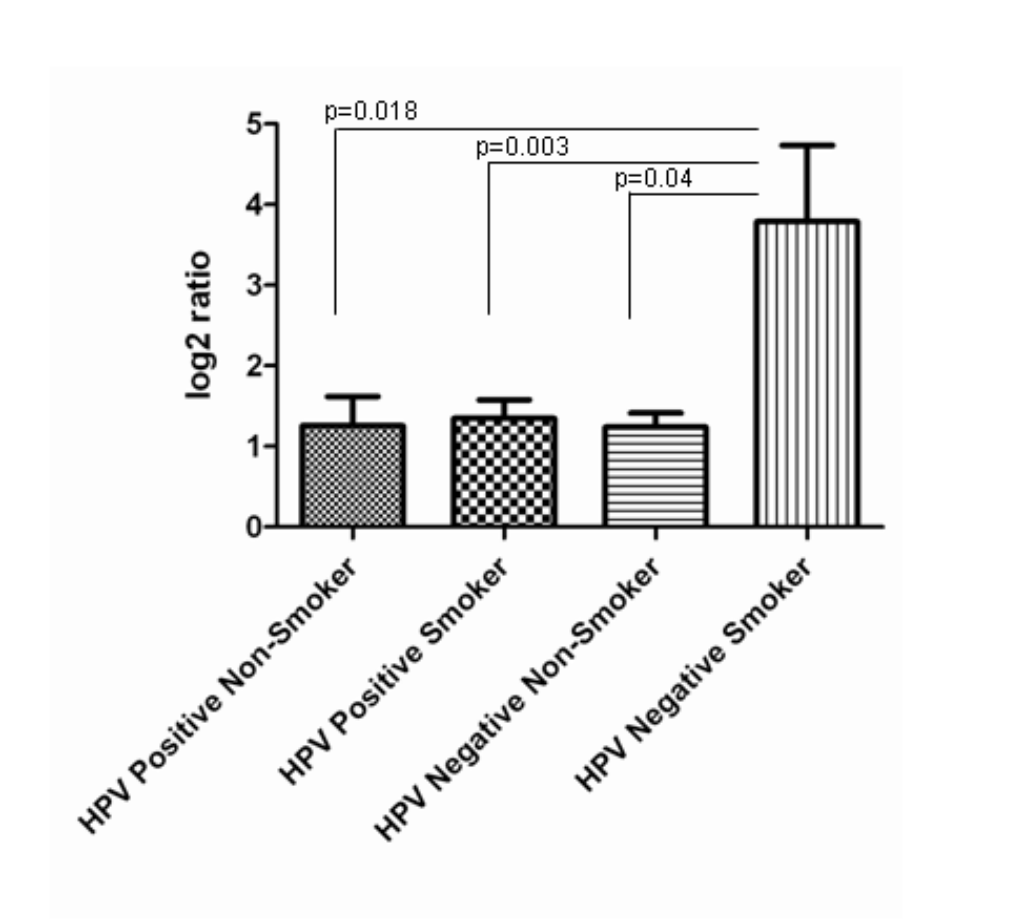
ATR ataxia telangiectasia and Rad3 related

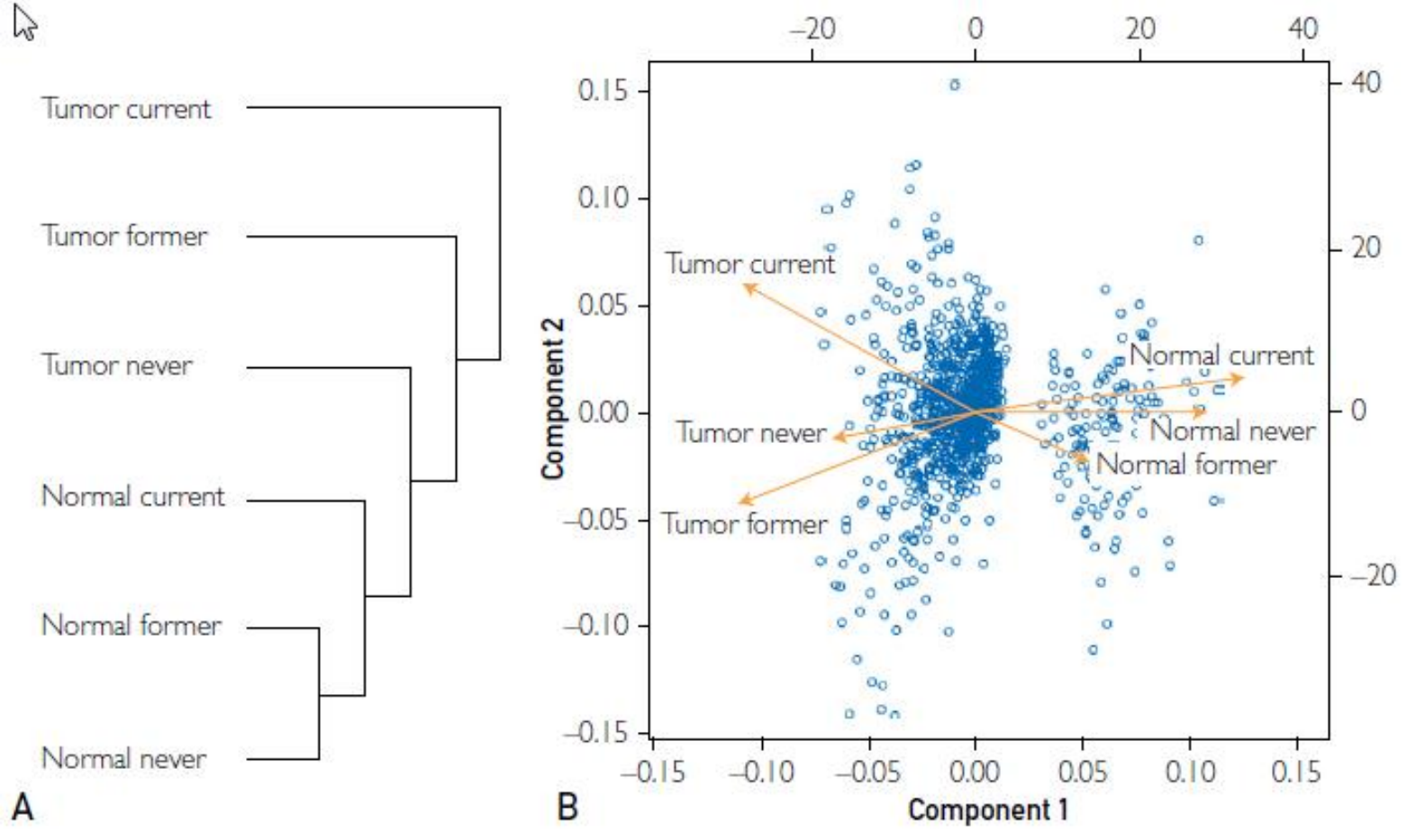


CHEK2

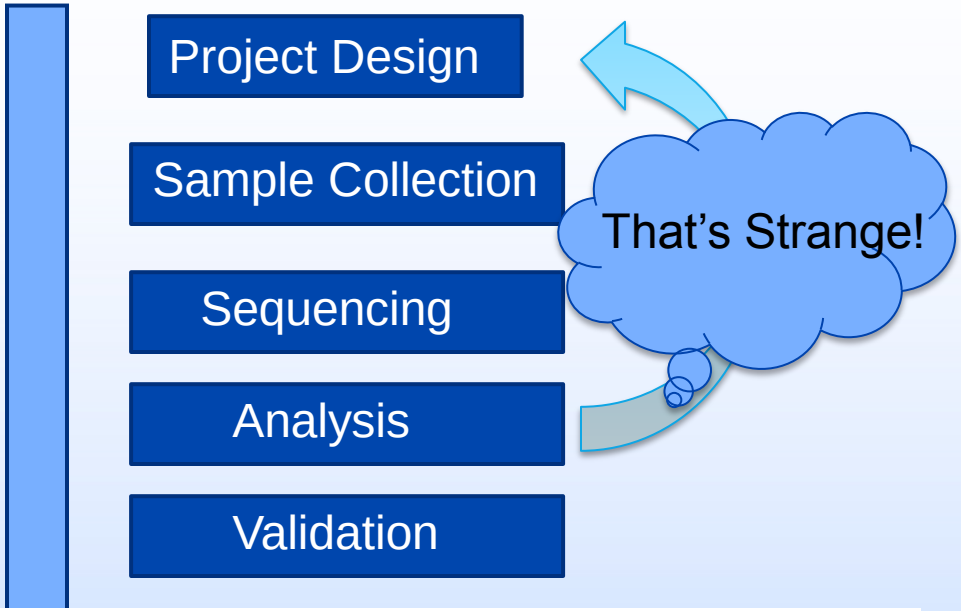

p=0.018
p=0.003
p=0.04
log2 ratio
5
4
3
2
1
0
HPV Positive Non-Smoker
HPV Positive Smoker
HPV Negative Non-Smoker
HPV Negative Smoker

Global Gene Expression

Smoking Status



The Analysis Process Inspires New Ideas



The Laryngoscope

Linking Expression of *FOXM1*, *CEP55* and *HELLS* to Tumorigenesis in Oropharyngeal Squamous Cell Carcinoma

Jeffrey R. Janus, MD*; Rebecca R. Laborde, PhD; Alexandra J. Greenberg, BA; Vivian W. Wang, PhD; Wei Wei, PhD; Anna Trier; Steven M. Olsen, MD; Eric J. Moore, MD; Kerry D. Olsen, MD; David I. Smith, PhD

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Additional Projects

HPV in oropharyngeal squamous cell carcinoma: Assessing virus presence in normal tissue and activity in cervical metastasis. In Press. *Laryngoscope*

Laborde RR, Janus JR, Olsen S, Wang VW, Garcia JJ, Graham R, Moore EJ, Kasperbauer JL, Price D, Olsen, Price M, Halling G and Smith DI. 2012.

Quantification of Human papillomavirus transcriptional activity by massively parallel sequencing and digital counting protocols in oropharyngeal squamous cell carcinoma. Submitted

Laborde RR, Olsen SM, Garcia JJ, Wang W, Olsen KD, Moore EJ, Kasperbauer JL, Tombers NM, Smith DI. 2012.

Overexpression of *CDC7* and other cell cycle regulatory genes is associated with tumor stage in oropharyngeal squamous cell carcinoma

Rebecca R. Laborde, Alexandra J. Greenberg, Jeffrey R. Janus, Vivian W Wang, Wei Wei, Steven Olsen, Eric J. Moore, Kerry D. Olsen, Nicole M. Tombers, David I. Smith

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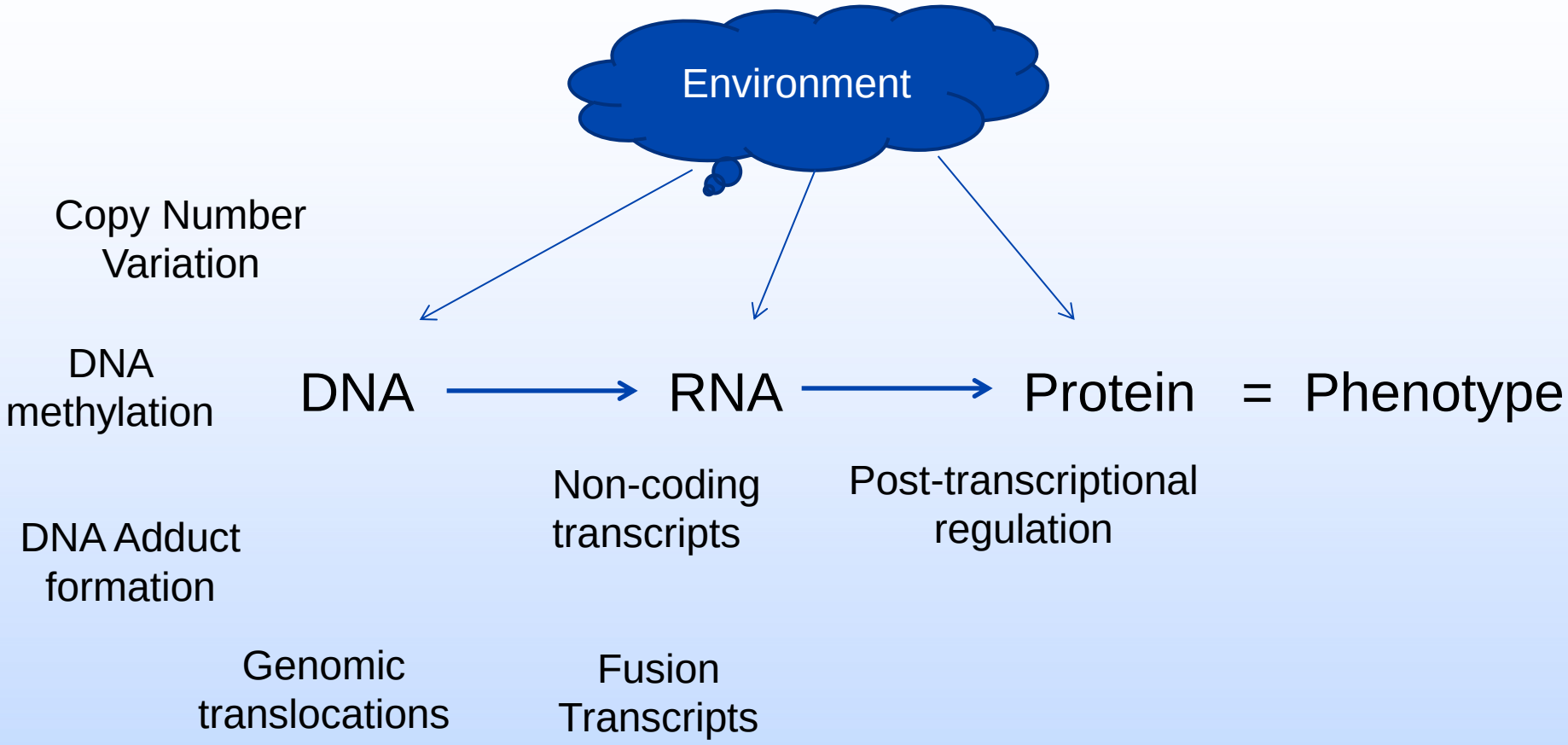
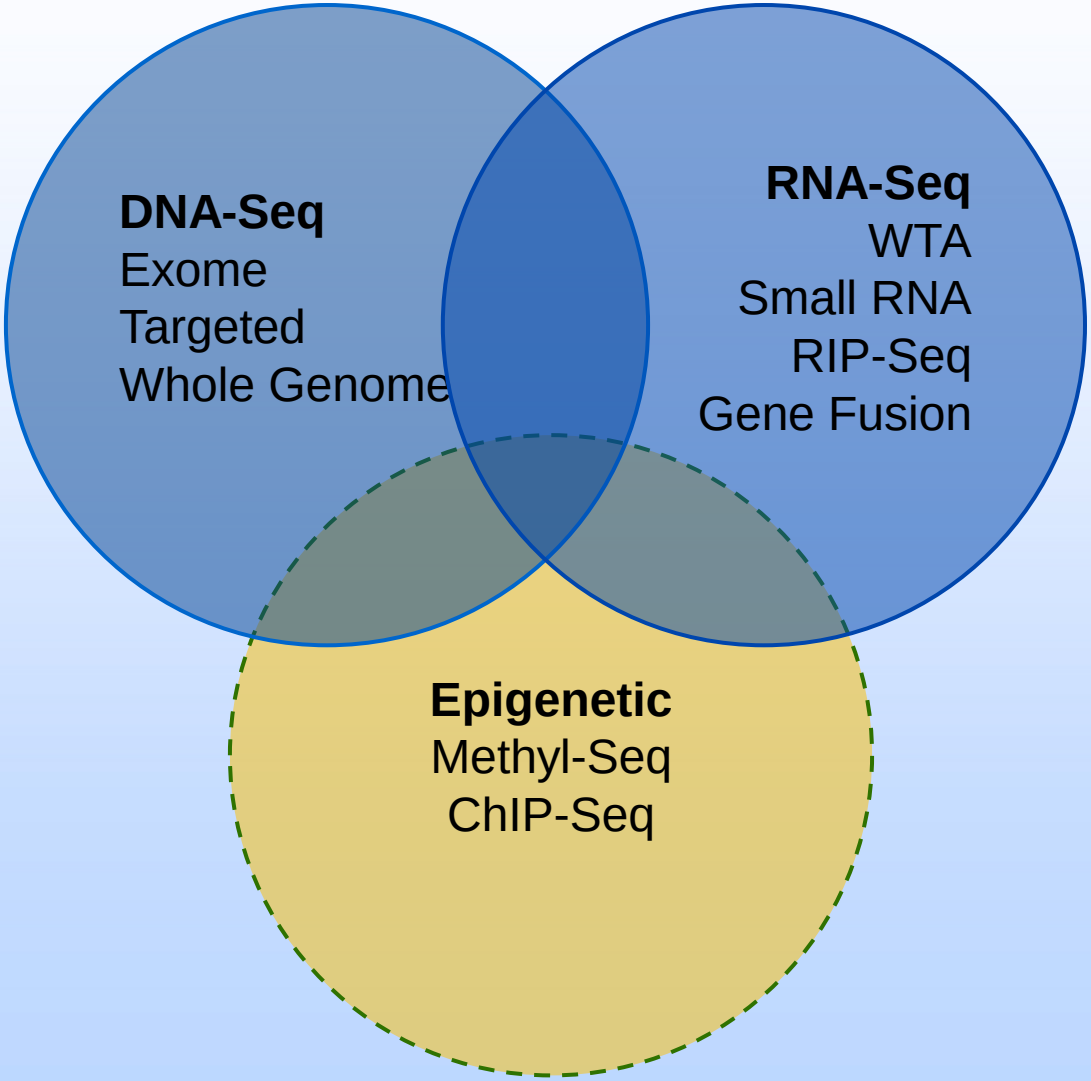
The power of multiple datasets



Generating Multiple Datasets Inter-Application Workflows



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Clinical Application: Saliva Screening Test

- Detection of disease by palpation of lesion and biopsy
  - Primary Disease
  - Recurrent Disease
- Clinicians in need of non-invasive screening tool
  - Saliva or swab biopsy

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What to target?

- HPV ?
  - Found in normal tissues
- Disease specific gene mutations
  - Develop a matrix detectable in shed epithelial cells
- Patient Tumor Specific gene mutations
  - Early detection of recurrent disease
  - Individualized medicine

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Exome Sequencing

Illumina TruSeq Library and Exome Enrichment kit

Sample	Age	Smoking	HPV
1	54	Current	Neg
2	46	Current	Neg
3	56	Current	Neg
4	69	Current	Neg
5	37	Never	Positive
6	57	Never	Positive
7	48	Never	Positive
8	54	Never	Positive

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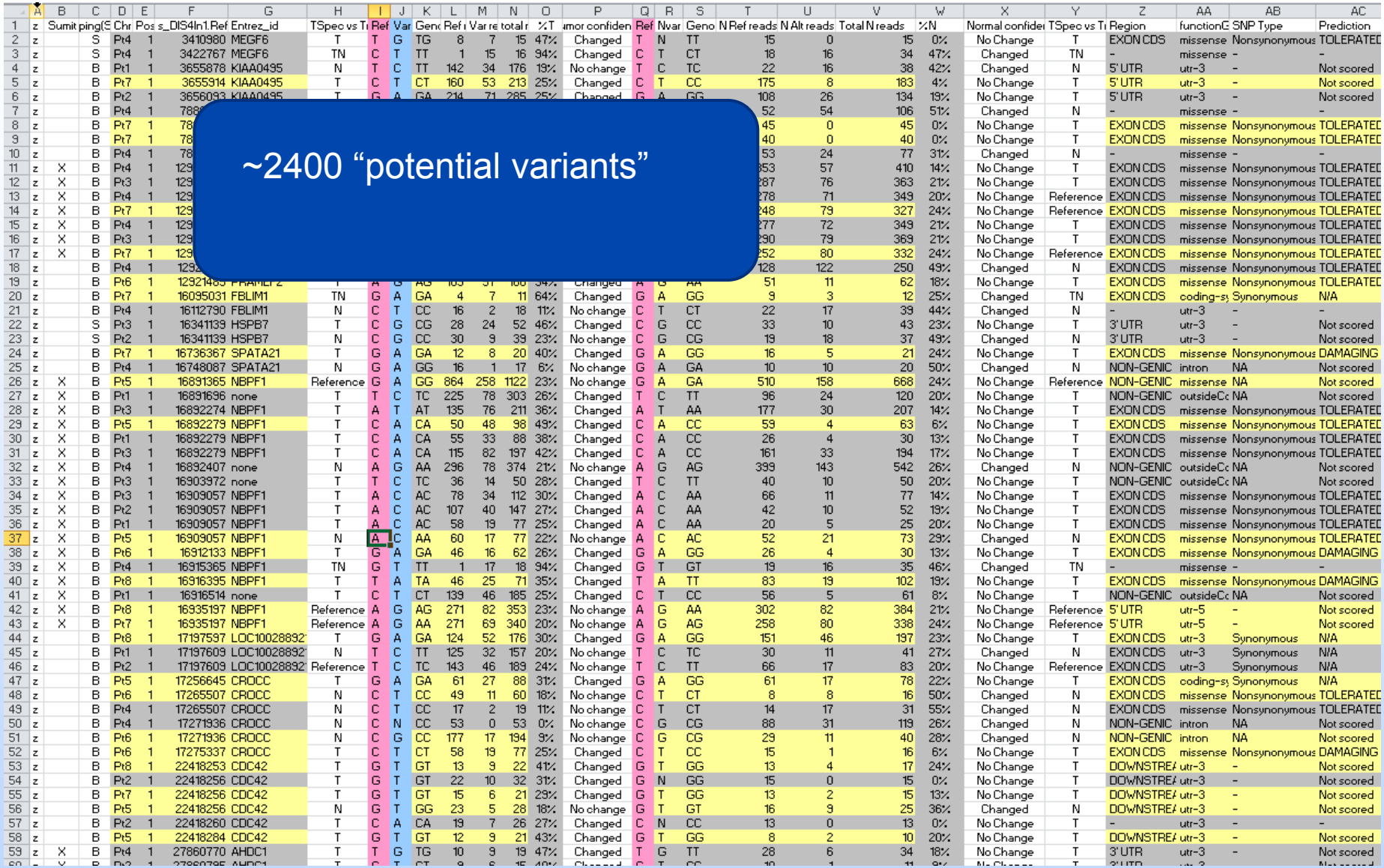
Exome data in Excel



	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U	V	W	X	Y	Z	AA	AB	AC
1	z	Sumit	ping(S	Chr	Pos	s_DIS4In1.Ref	Entrez_id	TSpec vs Ti	Ref	Var	Geno	Ref r	Var re	total r	%T	umor confiden	Ref	Nvar	Geno	N Ref reads	N Alt reads	Total N reads	%N	Normal confider	TSpec vs Ti	Region	functionG	SNP Type	Prediction
2	z		S	Pt4	1	3410980	MEGF6	T	T	G	TG	8	7	15	47%	Changed	T	N	TT	15	0	15	0%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
3	z		S	Pt4	1	3422767	MEGF6	TN	C	T	TT	1	15	16	94%	Changed	C	T	CT	18	16	34	47%	Changed	TN	-	missense	-	-
4	z		B	Pt1	1	3655878	KIAA0495	N	T	C	TT	142	34	176	19%	No change	T	C	TC	22	16	38	42%	Changed	N	5'UTR	utr-3	-	Not scored
5	z		B	Pt7	1	3655914	KIAA0495	T	C	T	CT	160	53	213	25%	Changed	C	T	CC	175	8	183	4%	No Change	T	5'UTR	utr-3	-	Not scored
6	z		B	Pt2	1	3656093	KIAA0495	T	G	A	GA	214	71	285	25%	Changed	G	A	GG	108	26	134	19%	No Change	T	5'UTR	utr-3	-	Not scored
7	z		B	Pt4	1	788														52	54	106	51%	Changed	N	-	missense	-	-
8	z		B	Pt7	1	78														45	0	45	0%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
9	z		B	Pt7	1	78														40	0	40	0%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
10	z		B	Pt4	1	78														53	24	77	31%	Changed	N	-	missense	-	-
11	z	X	B	Pt4	1	129														353	57	410	14%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
12	z	X	B	Pt3	1	129														287	76	363	21%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
13	z	X	B	Pt4	1	129														278	71	349	20%	No Change	Reference	EXON CDS	missense	Nonsynonymous	TOLERATED
14	z	X	B	Pt7	1	129														248	79	327	24%	No Change	Reference	EXON CDS	missense	Nonsynonymous	TOLERATED
15	z	X	B	Pt4	1	129														277	72	349	21%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
16	z	X	B	Pt3	1	129														290	79	369	21%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
17	z	X	B	Pt7	1	129														252	80	332	24%	No Change	Reference	EXON CDS	missense	Nonsynonymous	TOLERATED
18	z		B	Pt4	1	129														128	122	250	49%	Changed	N	EXON CDS	missense	Nonsynonymous	TOLERATED
19	z		B	Pt6	1	12921485	PHAMLP2	T	A	G	AG	103	51	166	34%	Changed	A	G	AA	51	11	62	18%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
20	z		B	Pt7	1	16095031	FBLIM1	TN	G	A	GA	4	7	11	64%	Changed	G	A	GG	9	3	12	25%	Changed	TN	EXON CDS	coding-sy	Synonymous	N/A
21	z		B	Pt4	1	16112790	FBLIM1	N	C	T	CC	16	2	18	11%	No change	C	T	CT	22	17	39	44%	Changed	N	-	utr-3	-	-
22	z		S	Pt3	1	16341139	HSPB7	T	C	G	CG	28	24	52	46%	Changed	C	G	CC	33	10	43	23%	No Change	T	3'UTR	utr-3	-	Not scored
23	z		S	Pt2	1	16341139	HSPB7	N	C	G	CC	30	9	39	23%	No change	C	G	CG	19	18	37	49%	Changed	N	3'UTR	utr-3	-	Not scored
24	z		B	Pt7	1	16736367	SPATA21	T	G	A	GA	12	8	20	40%	Changed	G	A	GG	16	5	21	24%	No Change	T	EXON CDS	missense	Nonsynonymous	DAMAGING
25	z		B	Pt4	1	16748087	SPATA21	N	G	A	GG	16	1	17	6%	No change	G	A	GA	10	10	20	50%	Changed	N	NON-GENIC	intron	NA	Not scored
26	z	X	B	Pt5	1	16891365	NBPF1	Reference	G	A	GG	864	258	1122	23%	No change	G	A	GA	510	158	668	24%	No Change	Reference	NON-GENIC	missense	NA	Not scored
27	z	X	B	Pt1	1	16891696	none	T	T	C	TC	225	78	303	26%	Changed	T	C	TT	96	24	120	20%	No Change	T	NON-GENIC	outsideCd	NA	Not scored
28	z	X	B	Pt3	1	16892274	NBPF1	T	A	T	AT	135	76	211	36%	Changed	A	T	AA	177	30	207	14%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
29	z	X	B	Pt5	1	16892279	NBPF1	T	C	A	CA	50	48	98	49%	Changed	C	A	CC	59	4	63	6%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
30	z	X	B	Pt1	1	16892279	NBPF1	T	C	A	CA	55	33	88	38%	Changed	C	A	CC	26	4	30	13%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
31	z	X	B	Pt3	1	16892279	NBPF1	T	C	A	CA	115	82	197	42%	Changed	C	A	CC	161	33	194	17%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
32	z	X	B	Pt4	1	16892407	none	N	A	G	AA	296	78	374	21%	No change	A	G	AG	399	143	542	26%	Changed	N	NON-GENIC	outsideCd	NA	Not scored
33	z	X	B	Pt3	1	16903972	none	T	T	C	TC	36	14	50	28%	Changed	T	C	TT	40	10	50	20%	No Change	T	NON-GENIC	outsideCd	NA	Not scored
34	z	X	B	Pt3	1	16909057	NBPF1	T	A	C	AC	78	34	112	30%	Changed	A	C	AA	66	11	77	14%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
35	z	X	B	Pt2	1	16909057	NBPF1	T	A	C	AC	107	40	147	27%	Changed	A	C	AA	42	10	52	19%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
36	z	X	B	Pt1	1	16909057	NBPF1	T	A	C	AC	58	19	77	25%	Changed	A	C	AA	20	5	25	20%	No Change	T	EXON CDS	missense	Nonsynonymous	TOLERATED
37	z	X	B	Pt5	1	16909057	NBPF1	N	A	C	AA	60	17	77	22%	No change	A	C	AC	52	21	73	29%	Changed	N	EXON CDS	missense	Nonsynonymous	TOLERATED
38	z	X	B	Pt6	1	16912133	NBPF1	T	G	A	GA	46	16	62	26%	Changed	G	A	GG	26	4	30	13%	No Change	T	EXON CDS	missense	Nonsynonymous	DAMAGING
39	z	X	B	Pt4	1	16915365	NBPF1	TN	G	T	TT	1	17	18	94%	Changed	G	T	GT	19	16	35	46%	Changed	TN	-	missense	-	-
40	z	X	B	Pt8	1	16916395	NBPF1	T	T	A	TA	46	25	71	35%	Changed	T	A	TT	83	19	102	19%	No Change	T	EXON CDS	missense	Nonsynonymous	DAMAGING
41	z	X	B	Pt1	1	16916514	none	T	C	T	CT	139	46	185	25%	Changed	C	T	CC	56	5	61	8%	No Change	T	NON-GENIC	outsideCd	NA	Not scored
42	z	X	B	Pt8	1	16935197	NBPF1	Reference	A	G	AG	271	82	353	23%	No change	A	G	AA	302	82	384	21%	No Change	Reference	5'UTR	utr-5	-	Not scored
43	z	X	B	Pt7	1	16935197	NBPF1	Reference	A	G	AA	271	69	340	20%	No change	A	G	AG	258	80	338	24%	No Change	Reference	5'UTR	utr-5	-	Not scored
44	z		B	Pt8	1	17197597	LOC10028932	T	G	A	GA	124	52	176	30%	Changed	G	A	GG	151	46	197	23%	No Change	T	EXON CDS	utr-3	Synonymous	N/A
45	z		B	Pt1	1	17197609	LOC10028932	N	T	C	TT	125	32	157	20%	No change	T	C	TC	30	11	41	27%	Changed	N	EXON CDS	utr-3	Synonymous	N/A
46	z		B	Pt2	1	17197609	LOC10028932	Reference	T	C	TC	143	46	189	24%	No change	T	C	TT	66	17	83	20%	No Change	Reference	EXON CDS	utr-3	Synonymous	N/A
47	z		B	Pt5	1	17256645	CROCC	T	G	A	GA	61	27	88	31%	Changed	G	A	GG	61	17	78	22%	No Change	T	EXON CDS	coding-sy	Synonymous	N/A
48	z		B	Pt6	1	17265507	CROCC	N	C	T	CC	49	11	60	18%	No change	C	T	CT	8	8	16	50%	Changed	N	EXON CDS	missense	Nonsynonymous	TOLERATED
49	z		B	Pt4	1	17265507	CROCC	N	C	T	CC	17	2	19	11%	No change	C	T	CT	14	17	31	55%	Changed	N	EXON CDS	missense	Nonsynonymous	TOLERATED
50	z		B	Pt4	1	17271936	CROCC	N	C	N	CC	53	0	53	0%	No change	C	G	CG	88	31	119	26%	Changed	N	NON-GENIC	intron	NA	Not scored
51	z		B	Pt6	1	17271936	CROCC	N	C	G	CC	177	17	194	9%	No change	C	G	CG	29	11	40	28%	Changed	N	NON-GENIC	intron	NA	Not scored
52	z		B	Pt6	1	17275337	CROCC	T	C	T	CT	58	19	77	25%	Changed	C	T	CC	15	1	16	6%	No Change	T	EXON CDS	missense	Nonsynonymous	DAMAGING
53	z		B	Pt8	1	22418253	CDC42	T	G	T	GT	13	9	22	41%	Changed	G	T	GG	13	4	17	24%	No Change	T	DOWNSTREA	utr-3	-	Not scored
54	z		B	Pt2	1	22418256	CDC42	T	G	T	GT	22	10	32	31%	Changed	G	N	GG	15	0	15	0%	No Change	T	DOWNSTREA	utr-3	-	Not scored
55	z		B	Pt7	1	22418256	CDC42	T	G	T	GT	15	6	21	29%	Changed	G	T	GG	13	2	15	13%	No Change	T	DOWNSTREA	utr-3	-	Not scored
56	z		B	Pt5	1	22418256	CDC42	N	G	T	GG	23	5	28	18%	No change	G	T	GT	16	9	25	36%	Changed	N	DOWNSTREA	utr-3	-	Not scored
57	z		B	Pt2	1	22418260	CDC42	T	C	A	CA	19	7	26	27%	Changed	C	N	CC	13	0	13	0%	No Change	T	-	utr-3	-	-
58	z		B	Pt5	1	22418284	CDC42	T	G	T	GT	12	9	21	43%	Changed	G	T	GG	8	2	10	20%	No Change	T	DOWNSTREA	utr-3	-	Not scored
59	z	X	B	Pt4	1	27860770	AHDC1	T	T	G	TG	10	9	19	47%	Changed	T	G	TT	28	6	34	18%	No Change	T	3'UTR	utr-3	-	Not scored
60	z	X	B	Pt2	1	27860785	AHDC1	T	C	T	CT	9	6	15	40%	Changed	C	T	CC	10	1	11	9%	No Change	T	3'UTR	utr-3	-	Not scored

~2400 “potential variants”

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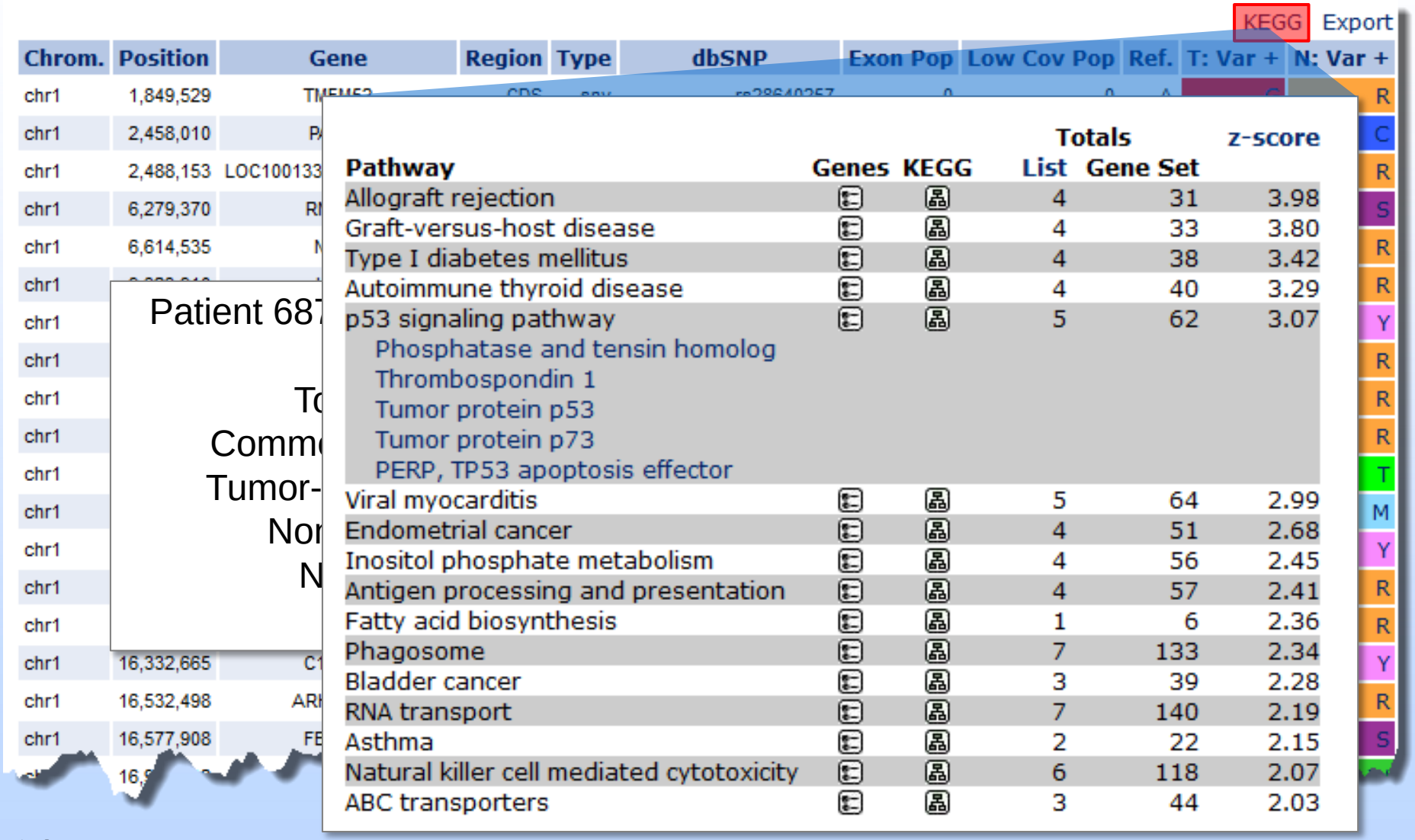
Exome



KEGG Export

Chrom.	Position	Gene	Region	Type	dbSNP	Exon Pop	Low Cov Pop	Ref.	T: Var +	N: Var +
chr1	1,849,529	TM	CDS	snv	rs28640257	0	0	A	C	R
chr1	2,458,010	P								C
chr1	2,488,153	LOC100133								R
chr1	6,279,370	R								S
chr1	6,614,535	N								R
chr1										R
chr1										Y
chr1										R
chr1										R
chr1										R
chr1										T
chr1										M
chr1										Y
chr1										R
chr1										R
chr1	16,332,665	C1								Y
chr1	16,532,498	ARI								R
chr1	16,577,908	FB								S
	16,9									

Patient 687

To
Comm
Tumor-
Nor
N

Pathway	Genes	KEGG	Totals List	Totals Gene Set	z-score
Allograft rejection			4	31	3.98
Graft-versus-host disease			4	33	3.80
Type I diabetes mellitus			4	38	3.42
Autoimmune thyroid disease			4	40	3.29
p53 signaling pathway			5	62	3.07
Phosphatase and tensin homolog					
Thrombospondin 1					
Tumor protein p53					
Tumor protein p73					
PERP, TP53 apoptosis effector					
Viral myocarditis			5	64	2.99
Endometrial cancer			4	51	2.68
Inositol phosphate metabolism			4	56	2.45
Antigen processing and presentation			4	57	2.41
Fatty acid biosynthesis			1	6	2.36
Phagosome			7	133	2.34
Bladder cancer			3	39	2.28
RNA transport			7	140	2.19
Asthma			2	22	2.15
Natural killer cell mediated cytotoxicity			6	118	2.07
ABC transporters			3	44	2.03

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Multi-Sample Comparisons VarScan

» Analysis Result File: **Multi-Sample Variant Report (SQLite)** — Number of Result

Can_score >4 + OR

Prediction possibly* -

T_663A: Effect non* -

Show: 20

All

Color Variants:

Search Reset

Expand All Collapse All

KEGG Export

Chrom.	Position	Gene	Region	Type	dbSNP	Exon Pop	Low Cov Pop	Ref.	N_663B: Var +	T_663A: Var +	Status	Pval	Ratio	Can_score	Cosmic	Prediction
chr1	1,019,753	C1orf159	CDS	snv	rs117262434	0	0	A	NC	R	Somatic	0.0371	0.82	17.13	0	possibly damaging
chr2	119,752,005	MARCO	CDS	snv	-	0	0	C	NC	Y	Somatic	0.0261	0	36.29	0	possibly damaging
chr2	180,835,580	CWC22	CDS	snv	-	0	0	A	NC	R	Somatic	2.7146e-06	1.11	29.21	0	possibly damaging
chr5	140,176,432	PCDHA@, PCDHA1, PCDHA2	CDS	snv	-	0	0	C	NC	Y	Somatic	1.3408e-07	1.15	40.44	0	possibly damaging
chr6	31,778,371	HSPA1L	CDS	snv	-	0	0	C	NC	Y	Somatic	9.8186e-09	0.64	31	0	possibly damaging
chr6	160,560,884	SLC22A1	CDS	snv	-	0	0	A	NC	W	Somatic	0.2089	0.25	16	0	possibly damaging
chr7	151,970,945	MLL3	CDS	snv	-	0	0	G	NC	K	Somatic	0.004	0.01	4.24	1	possibly damaging
chr8	24,771,356	NEFM	CDS	snv	-	0	0	T	NC	K	Somatic	0.193	0.5	26	0	possibly damaging
chr10	18,439,813	CACNB2	CDS	snv	-	0	0	C	NC	Y	Somatic	0.0085	0.48	18.5	0	possibly damaging
chr11	64,599,025	CDC42BPG	CDS	snv	-	0	0	C	NC	Y	Somatic	0.0433	2.33	41	0	possibly damaging
chr12	108,004,005	BTBD11	CDS	snv	-	0	0	C	NC	Y	Somatic	0.0086	0.33	26	0	possibly damaging
chr15	51,766,636	DMXL2	CDS	snv	-	0	0	G	NC	R	Somatic	0.0024	1.2	43.42	0	possibly damaging
chr15	91,436,551	FES	CDS	snv	-	0	0	A	R	R	Germline	1	0.17	4.13	0	possibly damaging
chr16	57,758,732	CCDC135	CDS	snv	rs2923144	5	0	C	M	M	Germline	1	0	4.19	0	possibly damaging
chr17	77,111,776	hCG_1776007	CDS	snv	-	0	0	C	NC	S	Somatic	0.0365	2	34.33	0	possibly damaging
chrX	24,073,777	EIF2S3	CDS	snv	-	0	0	C	NC	M	Somatic	0.0134	0.88	47.67	0	possibly damaging
chrX	77,913,338	ZCCHC5	CDS	snv	-	0	0	G	NC	K	Somatic	0.1833	1.2	19.18	0	possibly damaging

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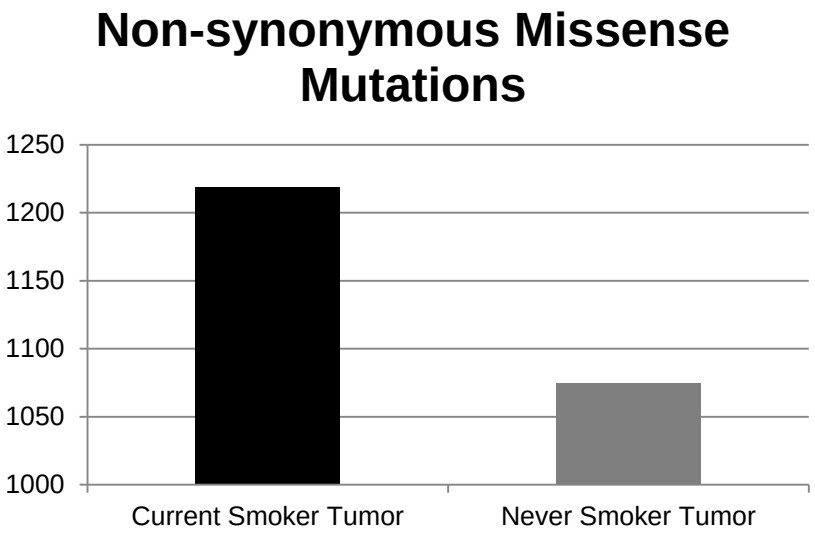
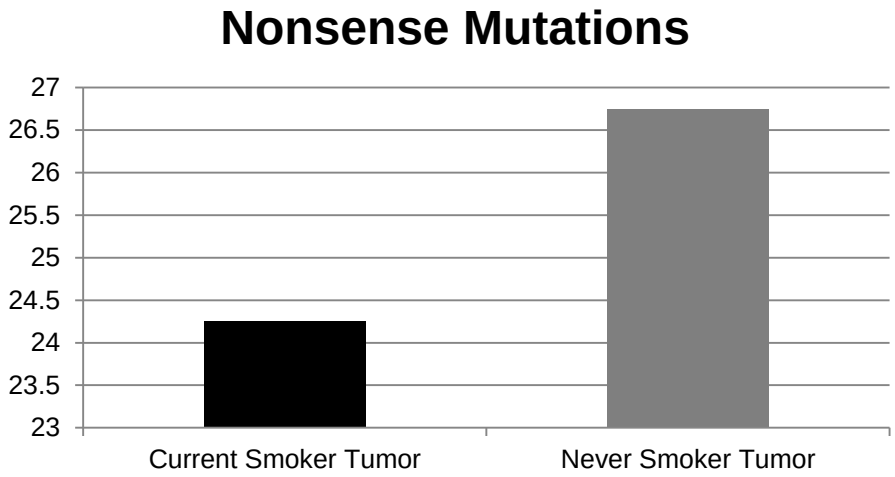
Variant Patterns in Head and Neck Cancers





Greater % Tumor Specific Variants

Generate Gene Lists

chr_pos	Description	Read Depth	Ref Frac	Alt Frac	Score	Gene	Change
chr2_97815072	G113, T19	132	0.856	0.144	171.29	ANKRD36	E359*
chr10_116930795	G80, T33	113	0.708	0.292	787.56	ATRNL1	E365*
chr8_2857565	G60, T18	78	0.769	0.231	374.29	CSMD1	C2706*
chr4_153251953	C70, T10	80	0.875	0.125	91.76	FBXW7	W351*
chr4_44680768	A3, G15	18	0.833	0.167	24.37	GUF1	W43*
chr5_23524525	C80, T16	96	0.833	0.167	248.97	PRDM9	R345*
chr10_897207441	C120, T15	135	0.889	0.111	91.07	PTEN	Q298*
chr11_678151915	G86, T12	98	0.878	0.122	117.71	TCIRG1	E463*
chr1_43149102	A59, G75	134	0.56	0.44	1664.78	YBX1	W65*

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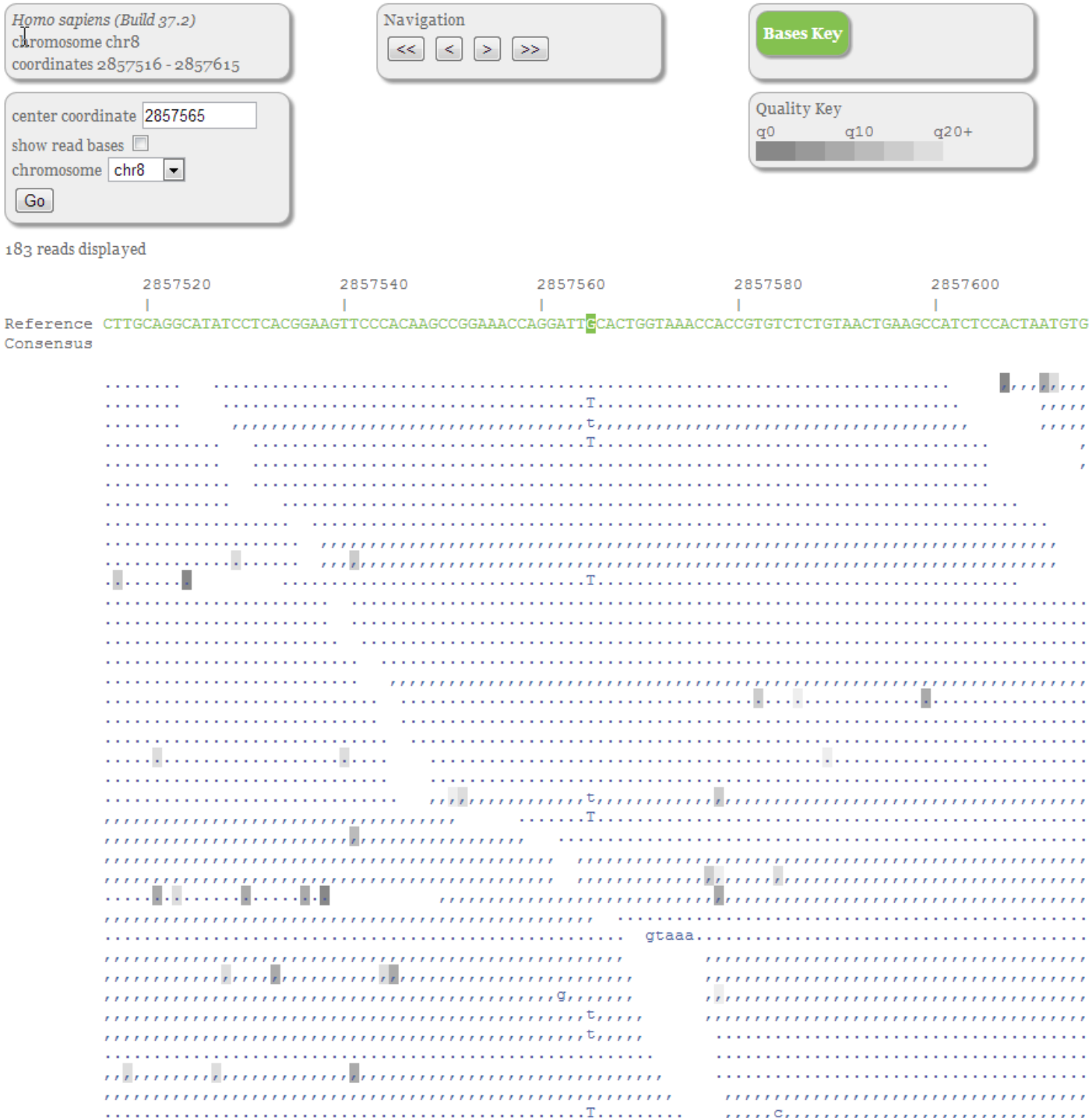
Homo sapiens (Build 37.2)
chromosome chr8
coordinates 2857516 - 2857615
Navigation
<< < > >>
Bases Key
center coordinate 2857565
show read bases
chromosome chr8
Go
Quality Key
q0 q10 q20+
183 reads displayed
2857520 2857540 2857560 2857580 2857600
Reference CTTGCAGGCATATCCTCACGGAAGTTCCCACAAGCCGGAAACCAGGATTGCACTGGTAAACCACCGTGTCTCTGTAACTGAAGCCATCTCCACTAATGTG
Consensus

Comparing Exome with RNA-Seq

									463 RNA-Seq		463 Exome	
									T	N	T	N
Chrom.	Position	Gene	Region	Type	dbSNP	Exon Pop	Low Cov Pop	Ref.	6745: Var +	6746: Var +	6747: Var +	6748: Var +
chr1	6,278,414	RNF207	CDS	snv	rs709209	0	0	A	NC	R	R	NC
chr1	43,149,102	YBX1	CDS	snv	-	0	0	G	R	NC	R	NC
chr2	10,263,618	RRM2	CDS	snv	-	0	0	G	K	NC	K	NC
chr2	27,600,585	ZNF513	CDS	snv	-	0	0	C	M	NC	M	NC
chr2	219,268,052	CTDSP1	CDS	snv	-	0	0	A	R	NC	R	NC
chr5	34,821,902	RAI14	CDS	snv	-	0	0	G	R	NC	R	NC
chr5	177,638,968	AGXT2L2	CDS	snv	-	0	0	A	R	NC	R	NC
chr6	109,787,200	ZBTB24	CDS	snv	-	0	0	C	M	NC	M	NC
chr8	32,463,096	NRG1	CDS	snv	-	0	0	A	W	NC	W	NC
chr8	98,731,403	MTDH	CDS	snv	-	0	0	G	R	NC	R	NC
chr11	104,900,433	CASP1	CDS	snv	-	0	0	T	Y	NC	Y	NC
chr11	134,118,751	THYN1	CDS	snv	-	0	0	T	Y	NC	Y	NC
chr12	64,521,471	SRGAP1	CDS	snv	rs115771292	1	0	C	Y	NC	Y	NC
chr16	31,470,886	ARMC5	CDS	snv	-	0	0	T	NC	W	W	NC
chr17	7,577,105	TP53	CDS	snv	-	0	0	G	S	NC	S	NC
chr17	40,257,981	DHX58	CDS	snv	-	0	0	C	M	NC	M	NC
chr20	33,511,157	ACSS2	CDS	snv	-	0	0	G	R	NC	R	NC
chr21	43,708,079	ABCG1	CDS	snv	-	0	0	G	S	NC	S	NC
chr1	12,854,414	PRAMEF1	CDS	snv	rs1063769	0	0	G	ND	ND	R	NC
chr1	12,907,781	HNRNPCL1	CDS	snv	rs1737105, rs28441396	0	0	T	ND	NC	Y	NC
chr1	12,907,798	HNRNPCL1	CDS	snv	-	0	0	A	NC	NC	M	NC
chr1	12,907,802	HNRNPCL1	CDS	snv	-	0	0	C	T	T	Y	NC
chr1	12,907,803	HNRNPCL1	CDS	snv	-	0	0	C	NC	NC	Y	NC
chr1	24,407,877	MYOM3	CDS	snv	-	0	0	C	ND	NC	Y	NC
chr1	40,960,924	ZNF642	CDS	snv	-	0	0	G	NC	NC	S	NC
chr1	144,854,581	PDE4DIP	CDS	snv	rs78371650	0	0	T	NC	NC	Y	NC
chr1	152,276,626	FLG	CDS	snv	rs3126075	0	0	G	ND	ND	S	NC

← CASP1

← PRAMEF1

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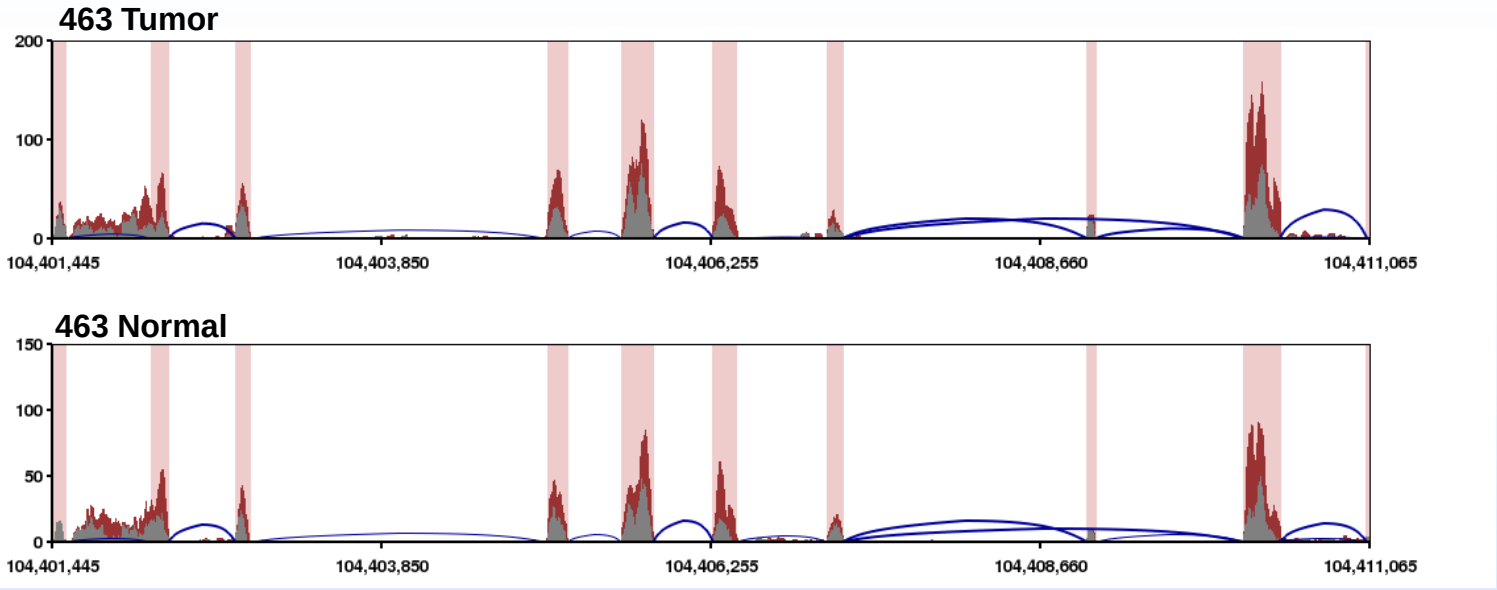
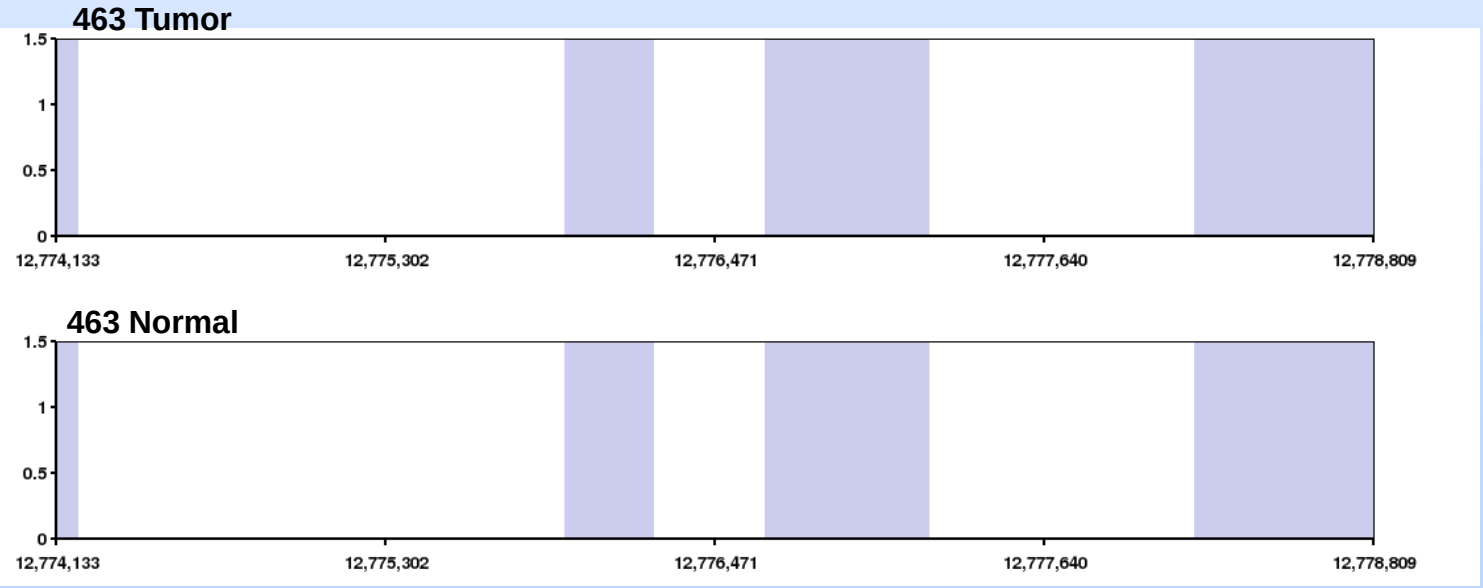
Exome with RNA-Seq



CASP1



PRAMEF1

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How to apply NGS to the clinics

- Identify clinically relevant targets in research samples
- Design workflows to interrogate these targets in large numbers of samples
- Store and manage data from large patient populations

- Analysis tools are key to all of these challenges

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Questions

PRESENTATIONS

Saturday, Mar 2, 1-2:00 pm

Breaking the Data Analysis Bottleneck:
Solutions That Work for RNA and Exome Sequencing
Presented by Rebecca Laborde, Mayo Clinic - SW1

Speaker @ Booth 522
Sun & Tue 10:30 - 11am

Monday, Mar 4, 11:00-12:30pm

Oyster Transcriptome Analysis by Next Gen Sequencing
Presented by Natalia Reyero, NHLBI - RG5

POSTERS

Sunday, Mar 3, 6:00-7:30 pm

Monday, Mar 4, 3:30-5:00 pm

#7 Identifying Mutations in Transcriptionally Active Regions of Genomes Using Next Generation Sequencing
Presented by Eric Olson, PerkinElmer

#11 What Does It Take to Identify the Signal from the Noise in Molecular Profiling of Tumors?
Presented by Eric Olson, PerkinElmer

award nominee

#119 Elucidating the Effects of the Deepwater Horizon Oil Spill on the Atlantic Oyster Using Global Transcriptome Analysis
Presented by Natalia Reyero, NHLBI

DEMONSTRATIONS

Sun 1pm / Mon 1:30pm

GeneSifter Analysis Edition at Booth 522

Sun 12:15 pm / Mon 3:30pm

GeneSifter LIMS at Booth 522

See a live demo of the easy to use bioinformatic software solutions designed by biologists for biologists and that were used in the talks and posters listed above.

PostShow Webinar - March 13, 10-11:00 am PST
Molecular Profiling of Tumors Using RNA and Exome Sequencing

PerkinElmer

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