

Genomics

Apply Genomics to Precision Medicine

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TRACO
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Outline

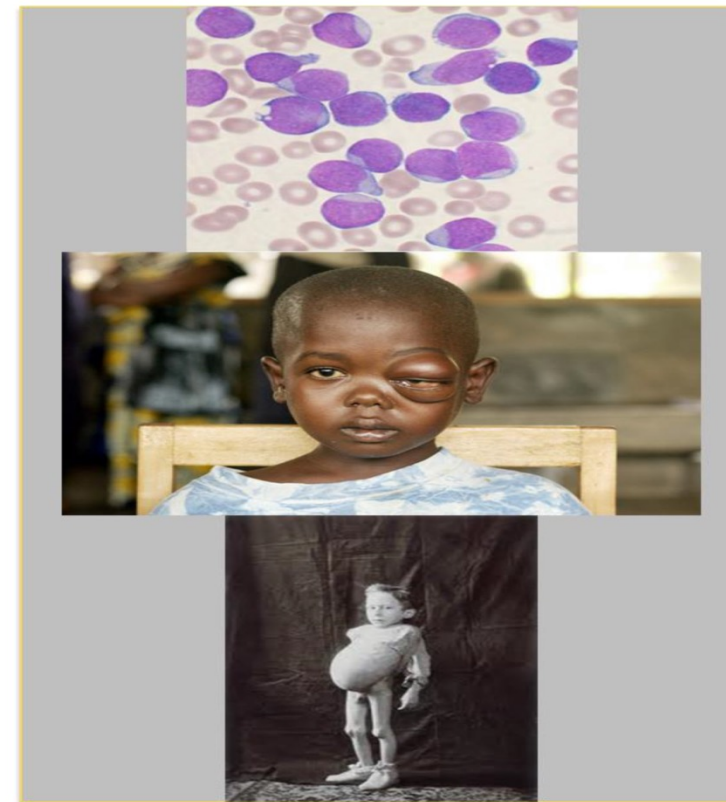
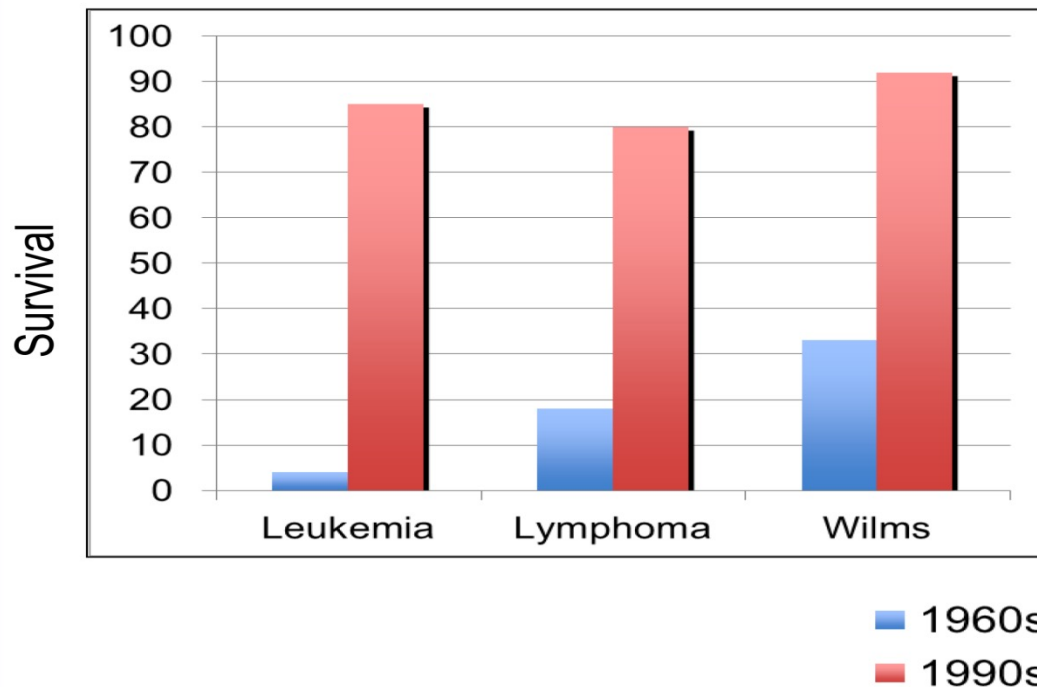
Outline

- **Success and Challenges of Treating Pediatric Cancers**
- **Genomics**
- **Next-generation Sequencing**
- **Application of next-generation sequencing:**
 - **Diagnosis**
 - **Identification of molecular target**
- **Precision Therapy**

Childhood cancer

National Cancer Institute

Childhood cancer: The beginning of a modern medical success story

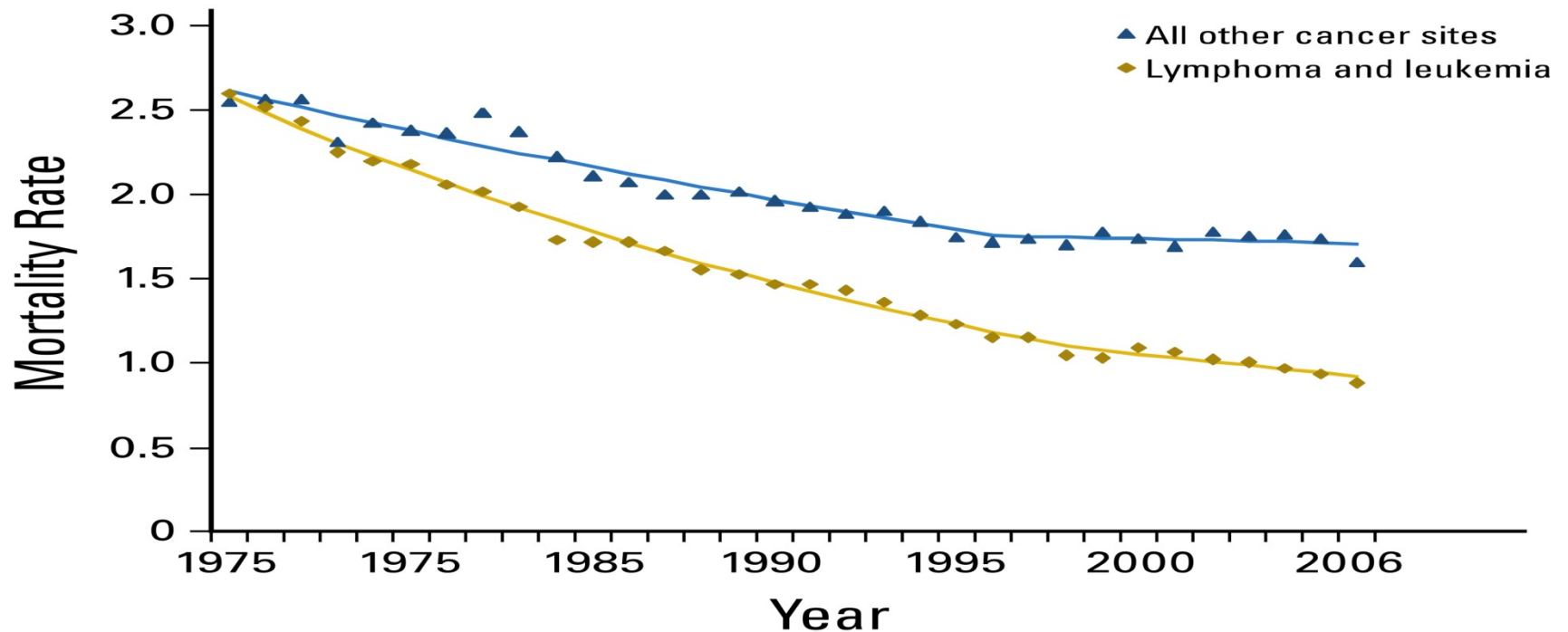


Courtesy: John Maris

Mortality rates

National Cancer Institute

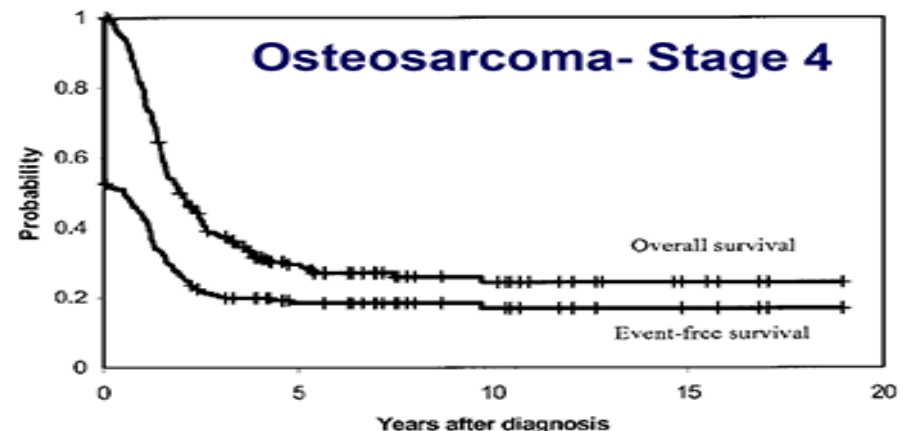
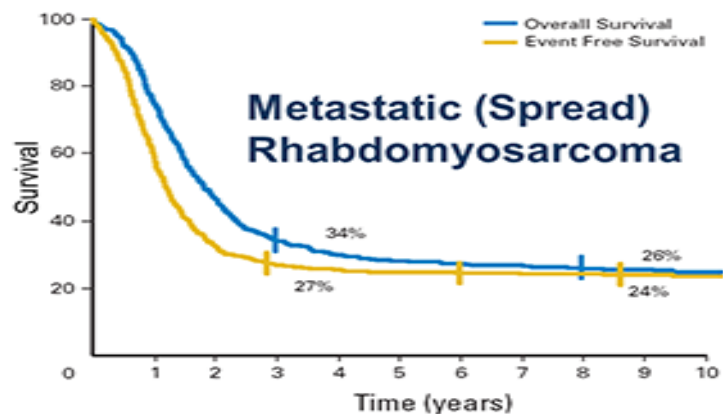
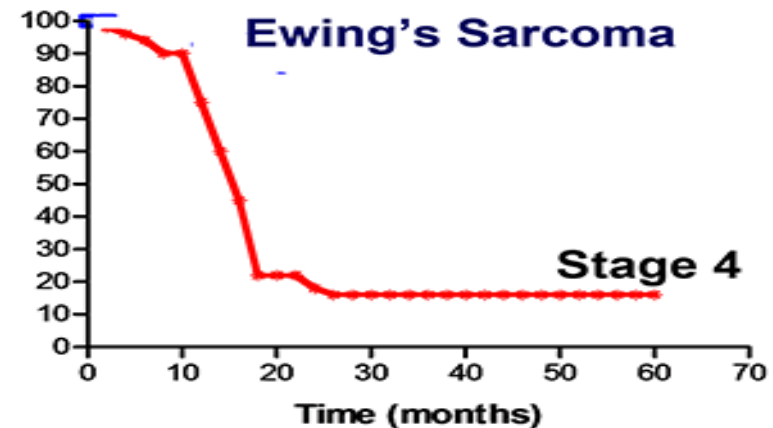
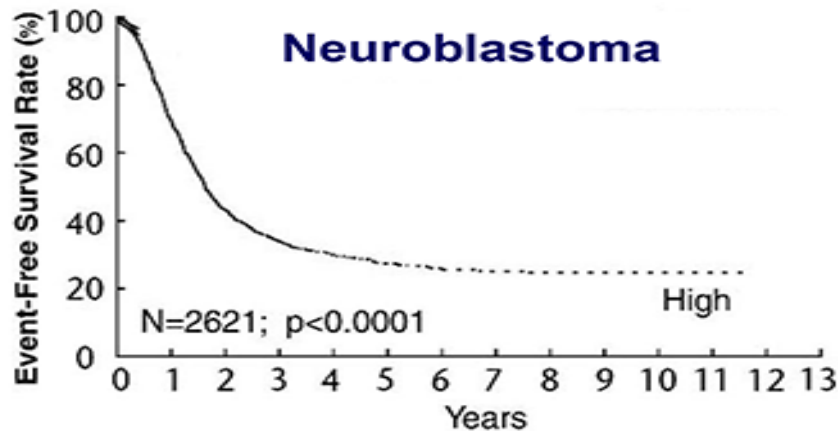
However in the past 16 years no improvement in mortality rates despite increased intensity of treatment



Courtesy: Malcolm Smith

Pediatric cancers

Metastatic, Recurrent, & Refractory Disease Remains Incurable



Gene expression

The dramatic consequences of gene expression in biology



Anise swallowtail, *Papilio zelicaon*

Same genome →
Different expression pattern
Different proteome
Different tissues
Different physiology

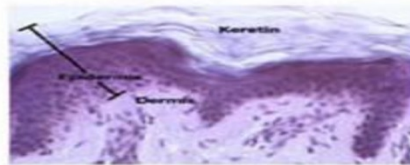


Gene expression

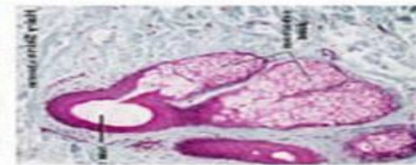
...but the complexity and diversity

Same genome or DNA →

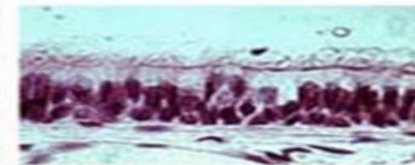
- Different expression pattern
- Different proteome
- Different tissues
- Different physiology



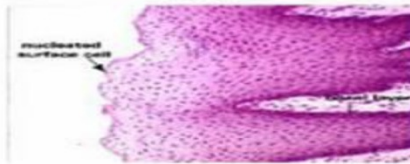
skin



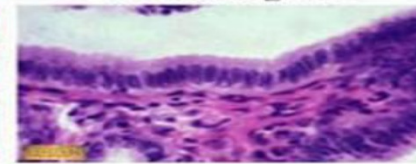
sebaceous gland



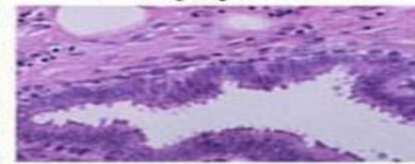
airway epithelium



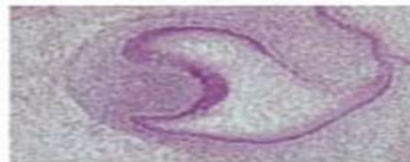
tongue



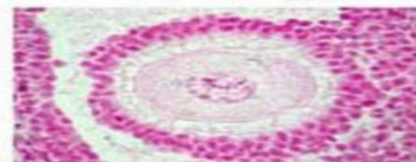
intestinal crypt



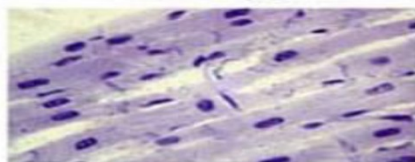
mammary gland



developing tooth



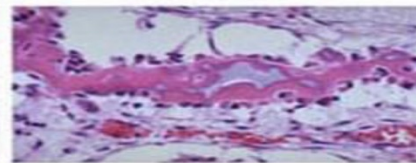
follicle



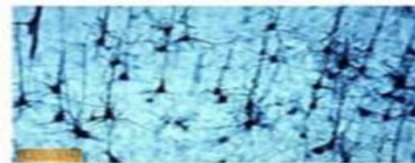
skeletal muscle



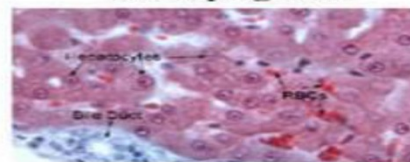
developing bone



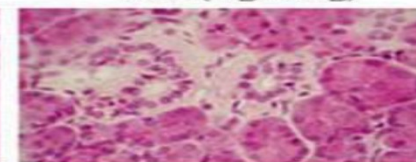
bone (high mag)



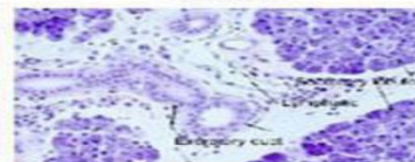
neuron



liver



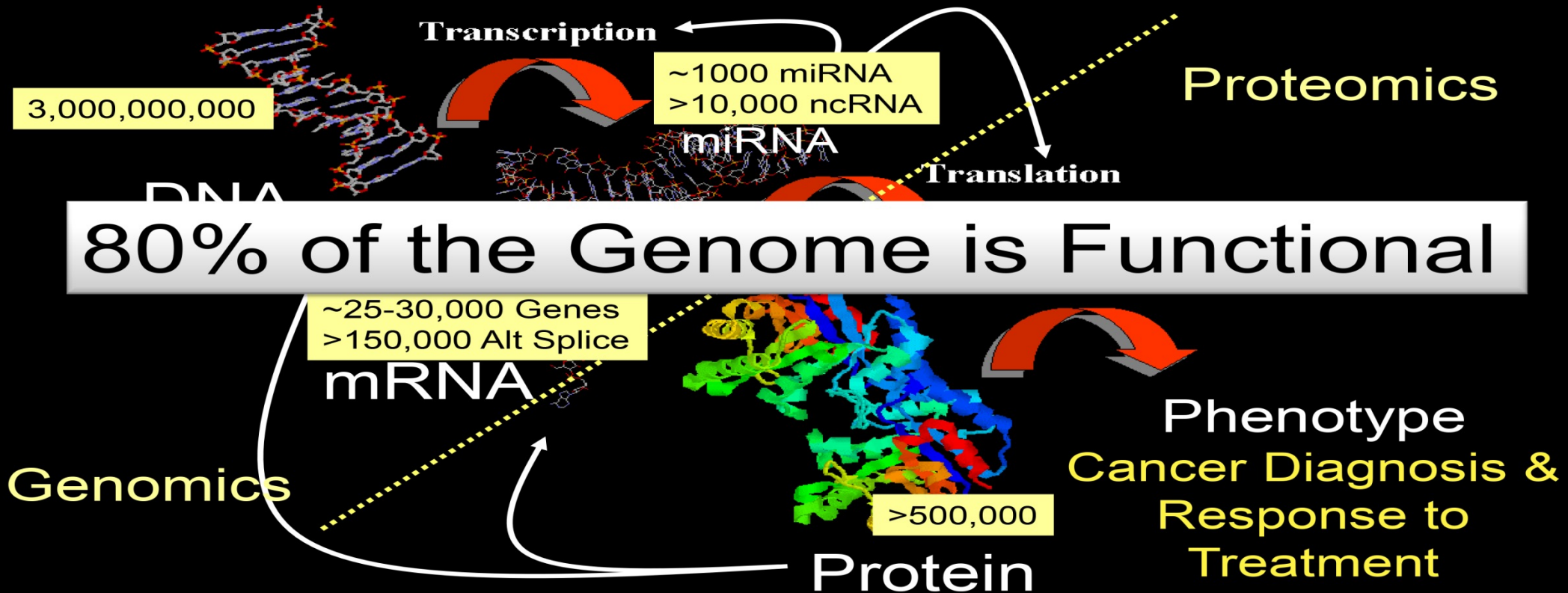
pancreas



parathyroid gland

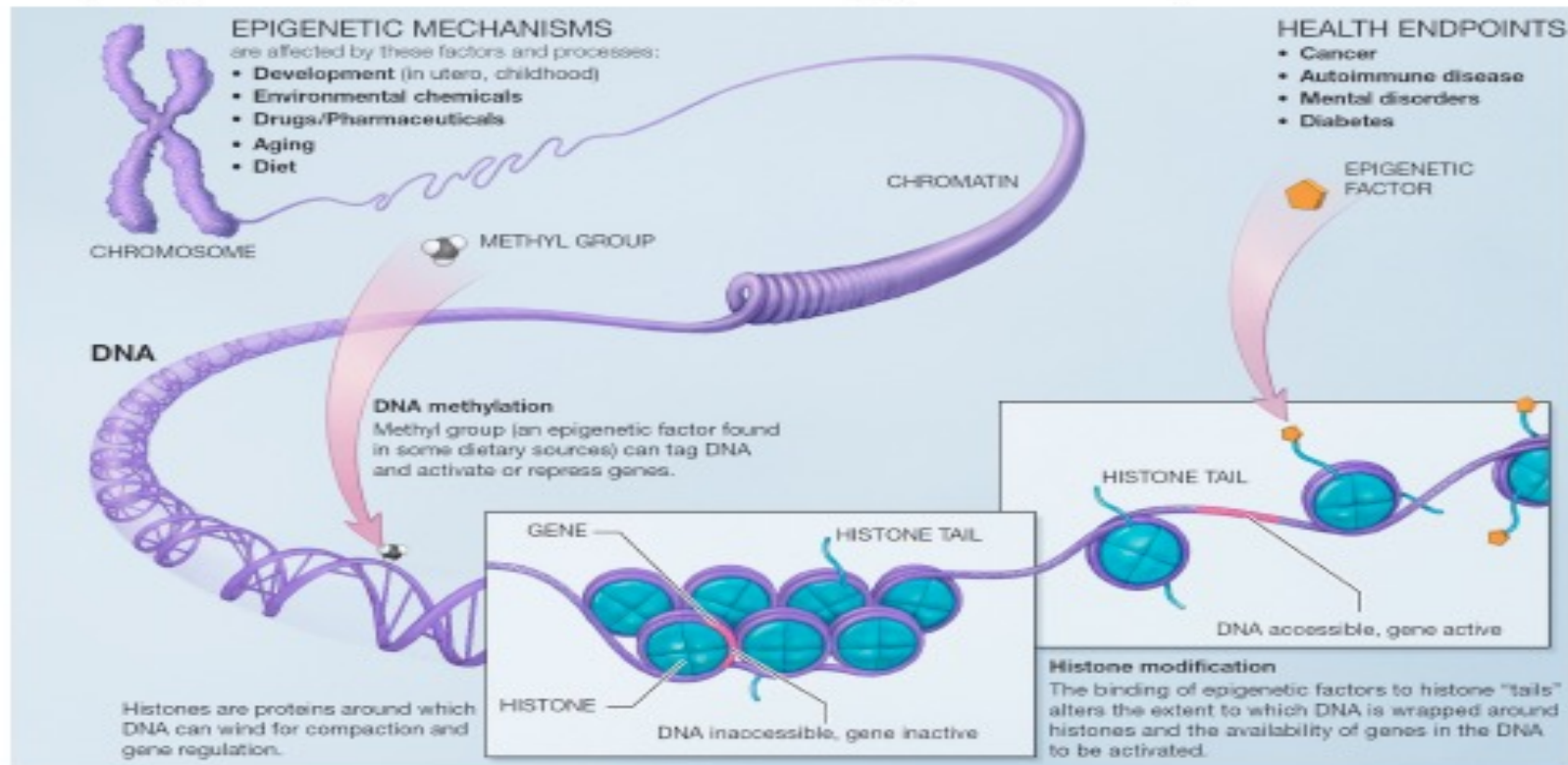
Gene expression

Biology is driven by the simultaneous expression of large numbers of genes acting in concert



Epigenetics

Epigenetics controls gene expression



Gene measurement

Challenge: how to measure/detect genes and their products in a massively parallel way?

- **High-throughput technologies**
- **Computational power**

Human genome



First generation tools

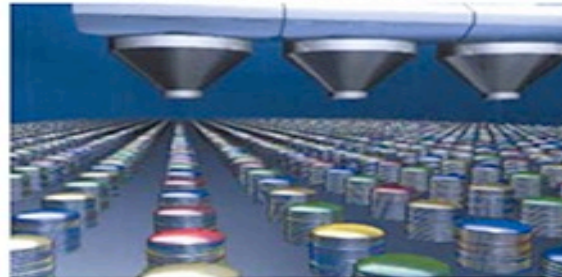
1st generation genomic tool: microarrays

Printing microarrays

Mechanical

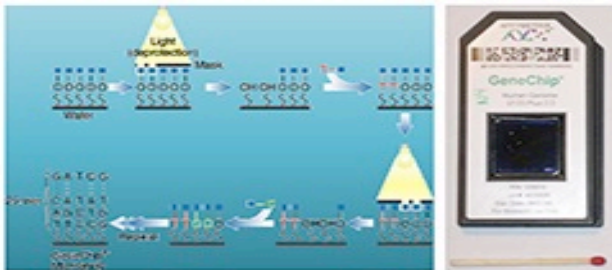


Electronic Piezo

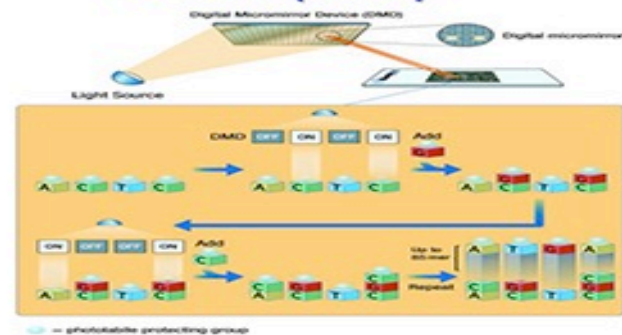


In-situ synthesis microarrays

Lithographic masks
and de-protection
through illumination

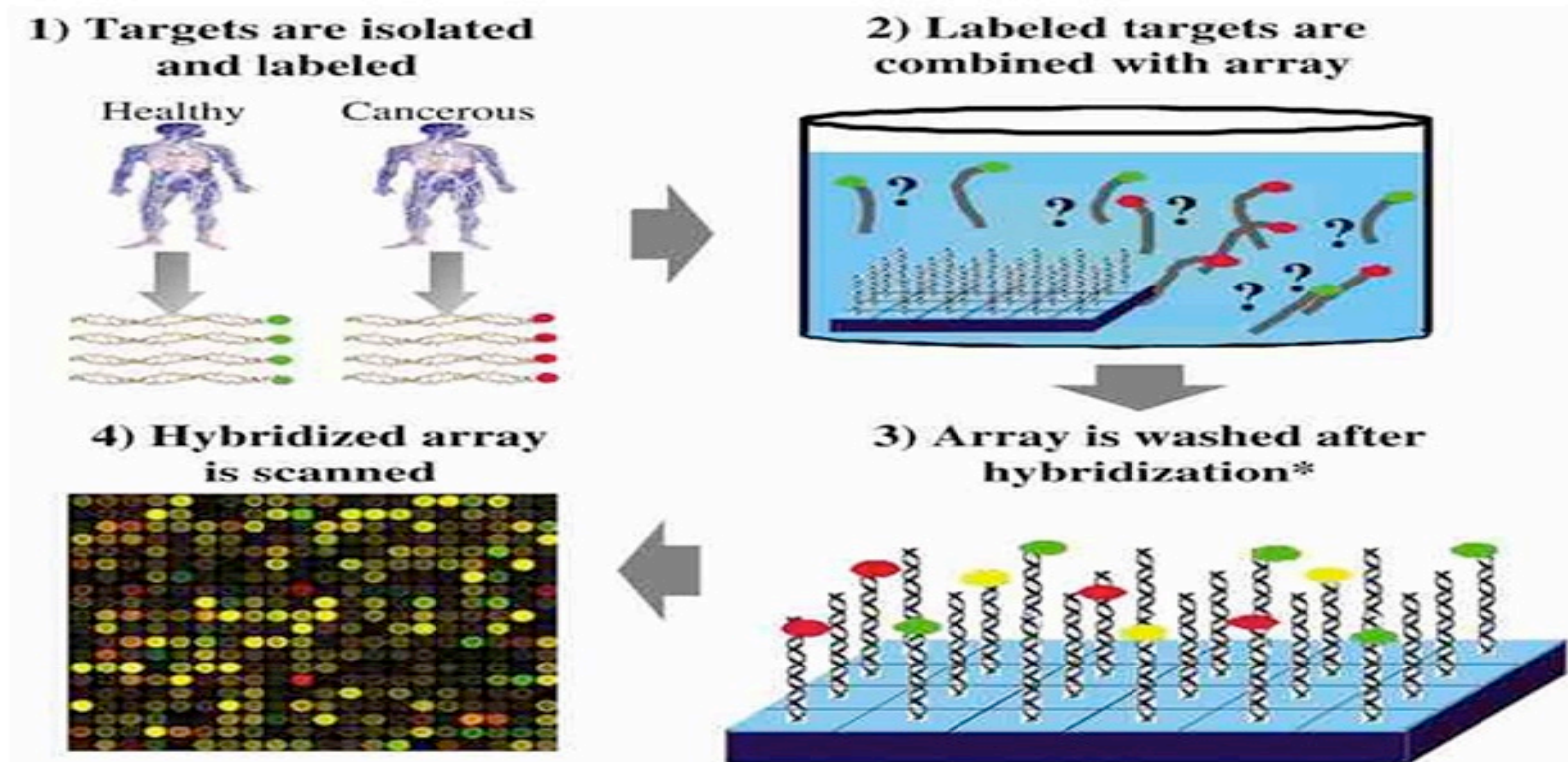


Digital micromirror device (DMD)



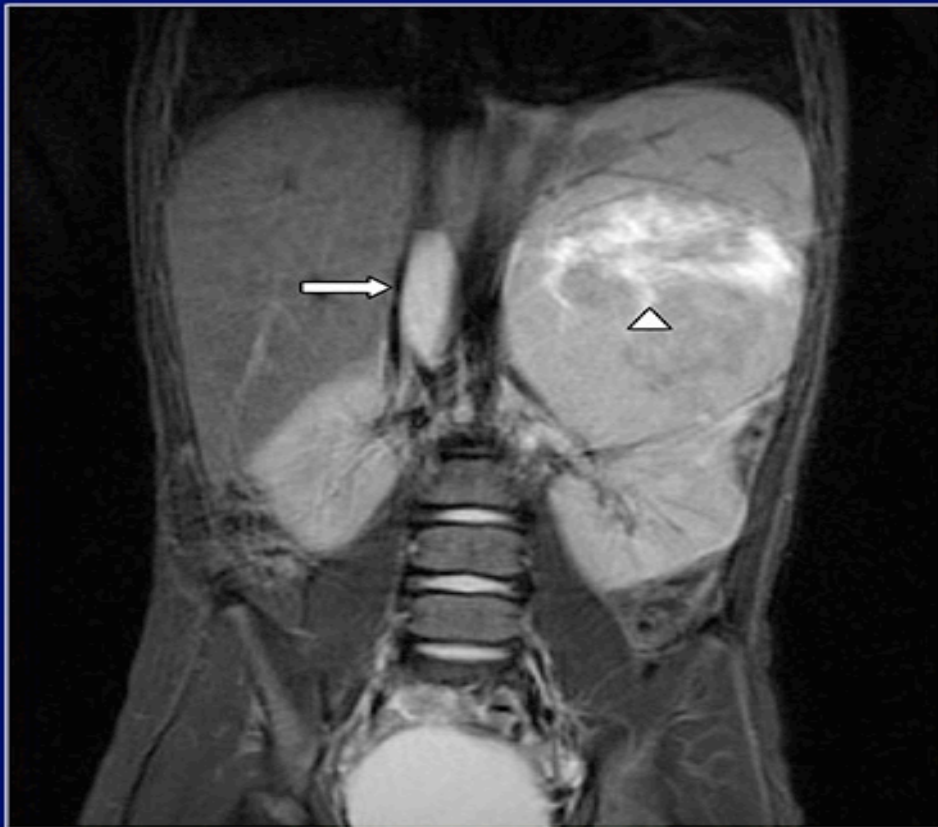
Microarrays

Microarrays – technologies of hybridization



Wilms tumor

MRI: 9 x 8 x 9 cm mass in upper pole left kidney, tumor in Left renal vein and inferior vena cava

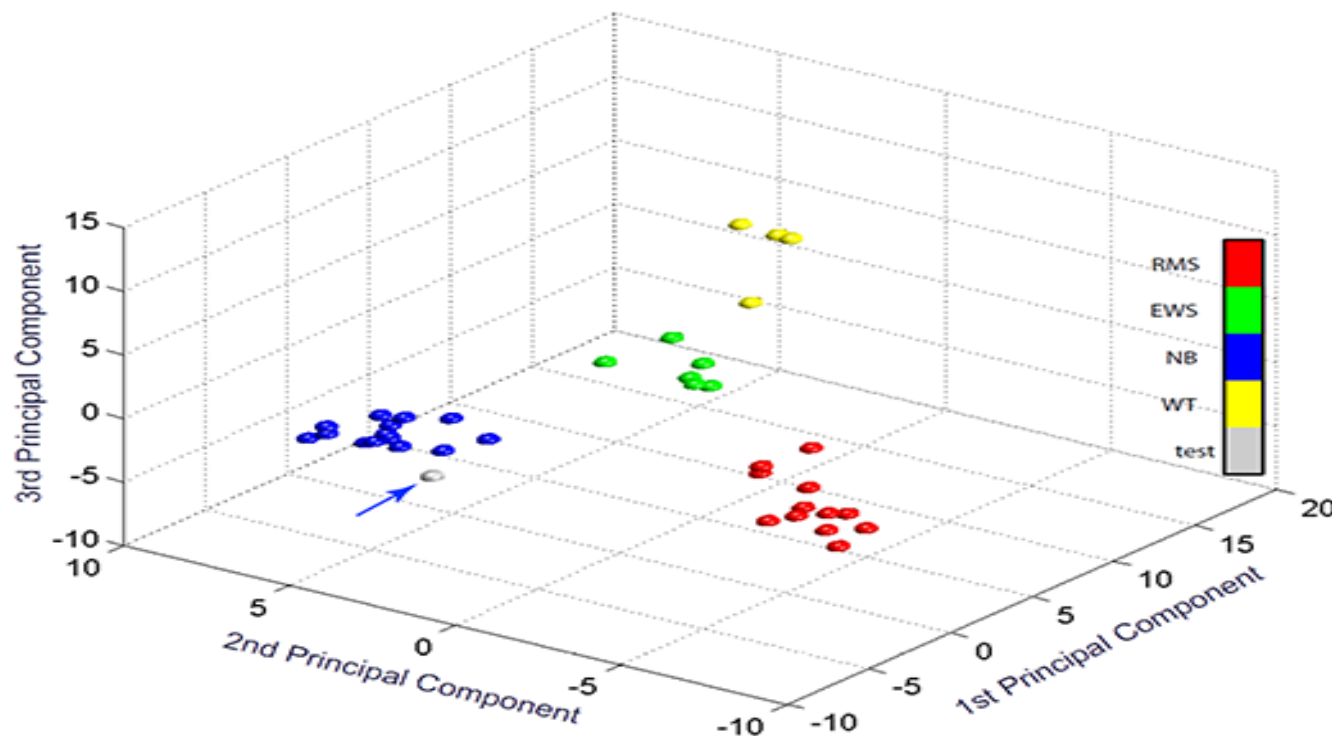


Initial diagnosis: Wilm's tumor



Cancer diagnosis

Diagnosis of cancers using gene expression profiles



Wilm's tumor

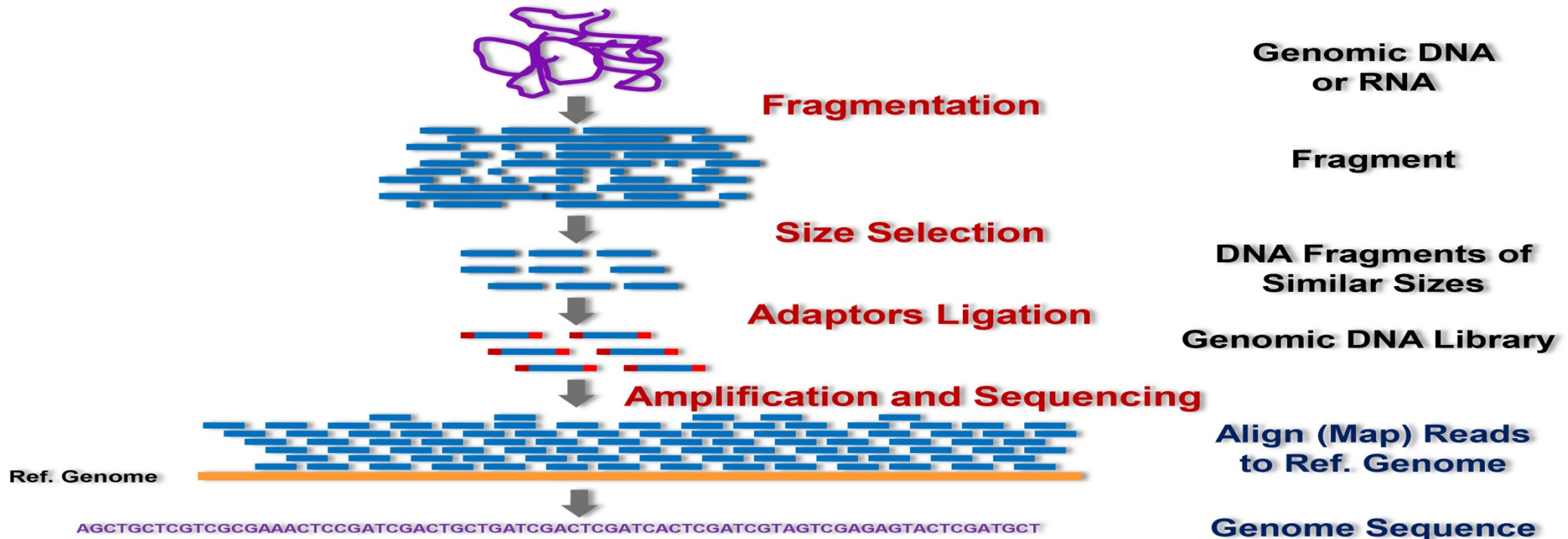


Neuroblastoma

- Patient was switched to high risk neuroblastoma treatment included stem cell transplant
- Doing well 1 yr after diagnosis

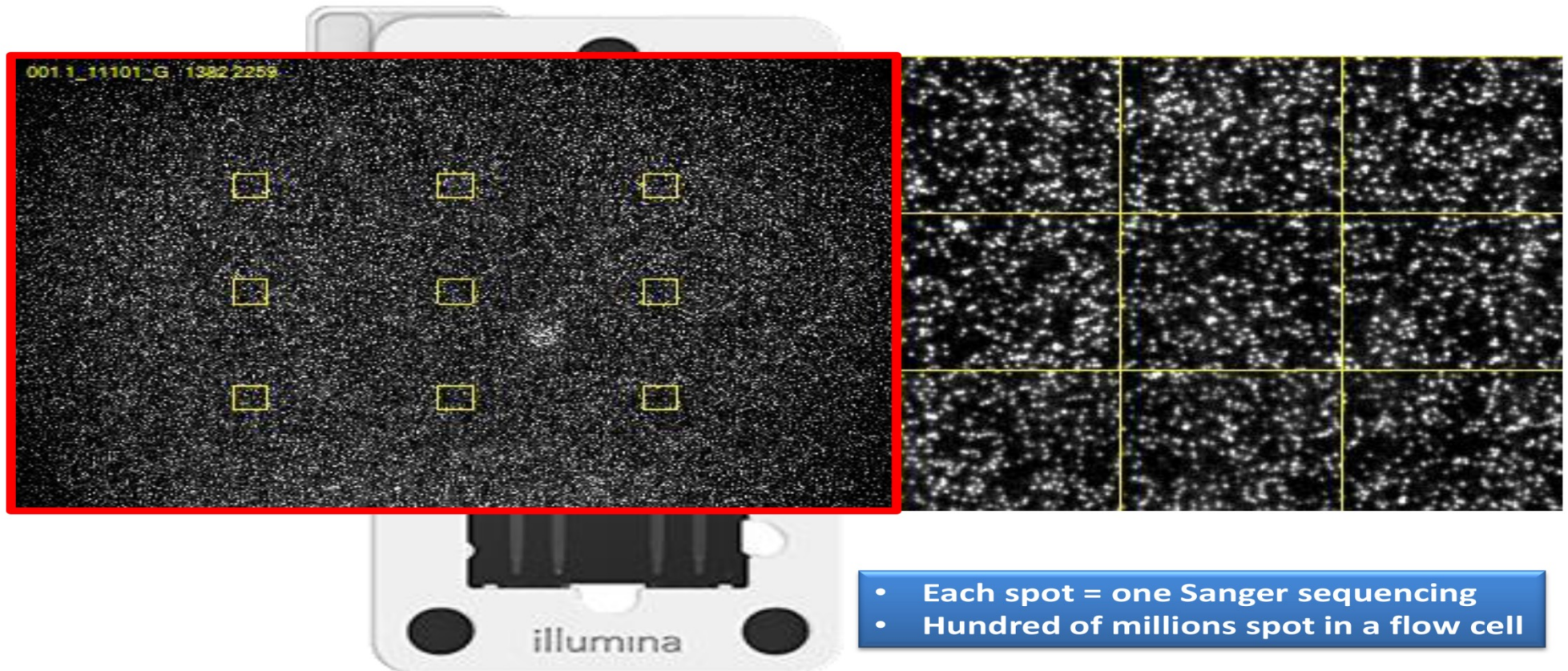
Next-generation sequencing

Next-Generation Sequencing



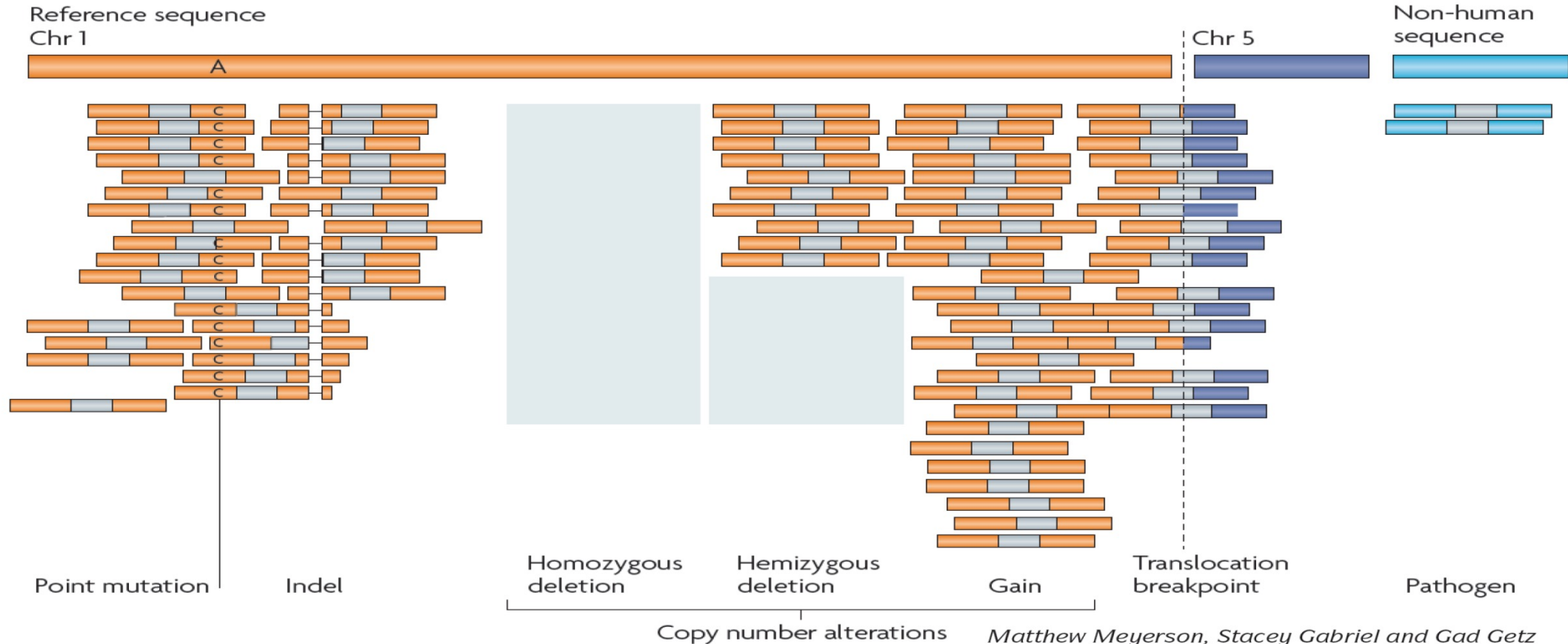
Massively Parallel Sequencing

Massively Parallel Sequencing



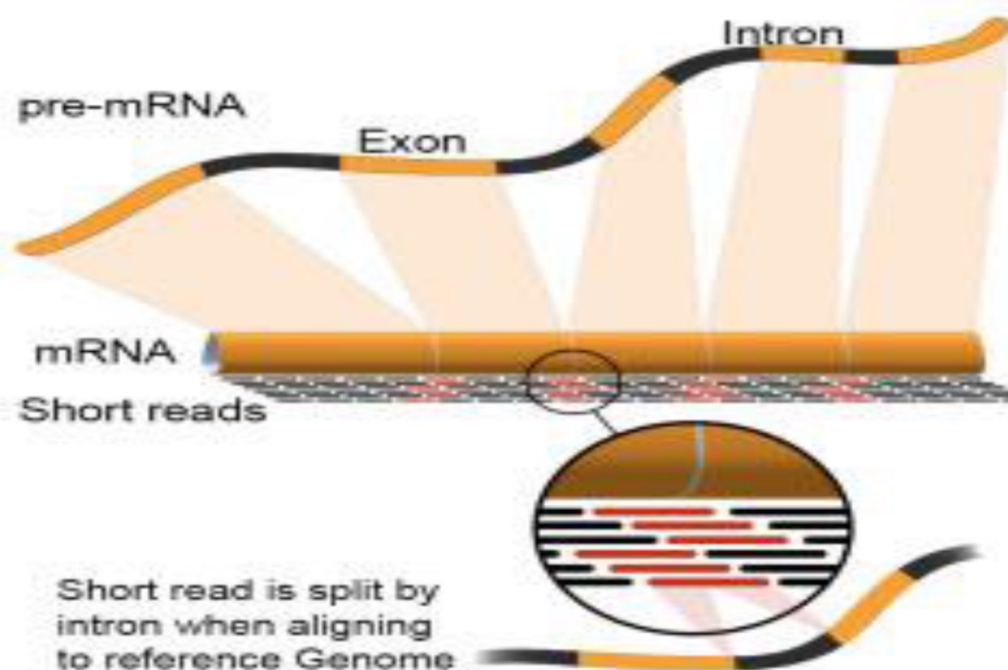
Genomic Alterations

Genomic alterations detected by DNA sequencing



Genomic Alterations

Genomic Alterations Detected by RNA Transcriptome Sequencing



- Digital Gene Expression
- Expressed Mutations
- Alternative Splicing Events
- Expressed Fusion Transcripts
- RNA editing
- Novel Transcripts
- Non-coding RNAs

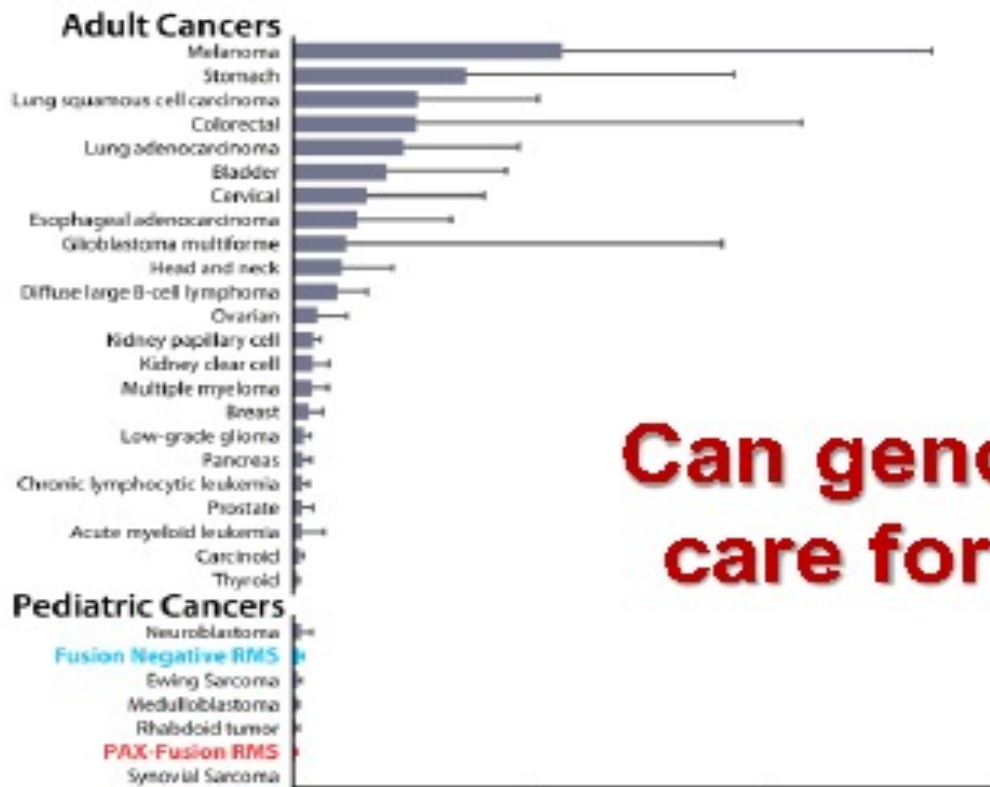
Next-generation sequencing

Next-generation sequencing: a platform for many applications to study genome and epigenome

- No need of prior knowledge for probe design as in microarrays
- Parallel sequencing at basepair resolution– massive-throughput
 - Then: *~13 years for the 1st human genome* using Sanger sequencing by 20 centers in 7 countries
 - Now: *multiple human genomes in 2 days* using a NGS sequencer
- A single platform for different kinds of genomic and epigenomic information
 - DNA and RNA sequencing
 - Genome modification, e.g. methylation
 - Chromatin accessibility, e.g. ATAC-seq
 - Chromatin 3D organization, e.g. Hi-C
 - Protein-DNA interaction, e.g. ChIP-seq

Pediatric cancer mutations

Pediatric Cancers Have A Low Number of Somatic and Actionable Mutations At Initial Diagnosis



Can genomics help clinical care for cancer patients?

Clinomics for precision medicine

Personalized Medicine and Imaging

Clinical
Cancer
Research

MultiDimensional ClinOmics for Precision Therapy of Children and Adolescent Young Adults with Relapsed and Refractory Cancer: A Report from the Center for Cancer Research

Wendy Chang^{1,2,3}, Andrew S. Brohl^{1,4}, Rajesh Patidar¹, Sivasish Sindiri¹, Jack F. Shern^{1,2}, Jun S. Wei¹, Young K. Song¹, Marielle E. Yohe^{1,2}, Berkley Gryder¹, Shile Zhang¹, Kathleen A. Calzone⁵, Nityashree Shivaprasad¹, Xinyu Wen¹, Thomas C. Badgett^{1,6}, Markku Miettinen⁷, Kip R. Hartman^{8,9}, James C. League-Pascual^{2,8}, Toby N. Trahair¹⁰, Brigitte C. Widemann², Melinda S. Merchant², Rosandra N. Kaplan², Jimmy C. Lin¹, and Javed Khan¹

Clin Cancer Res. May 2016

Protocol Number: 10-C-0086

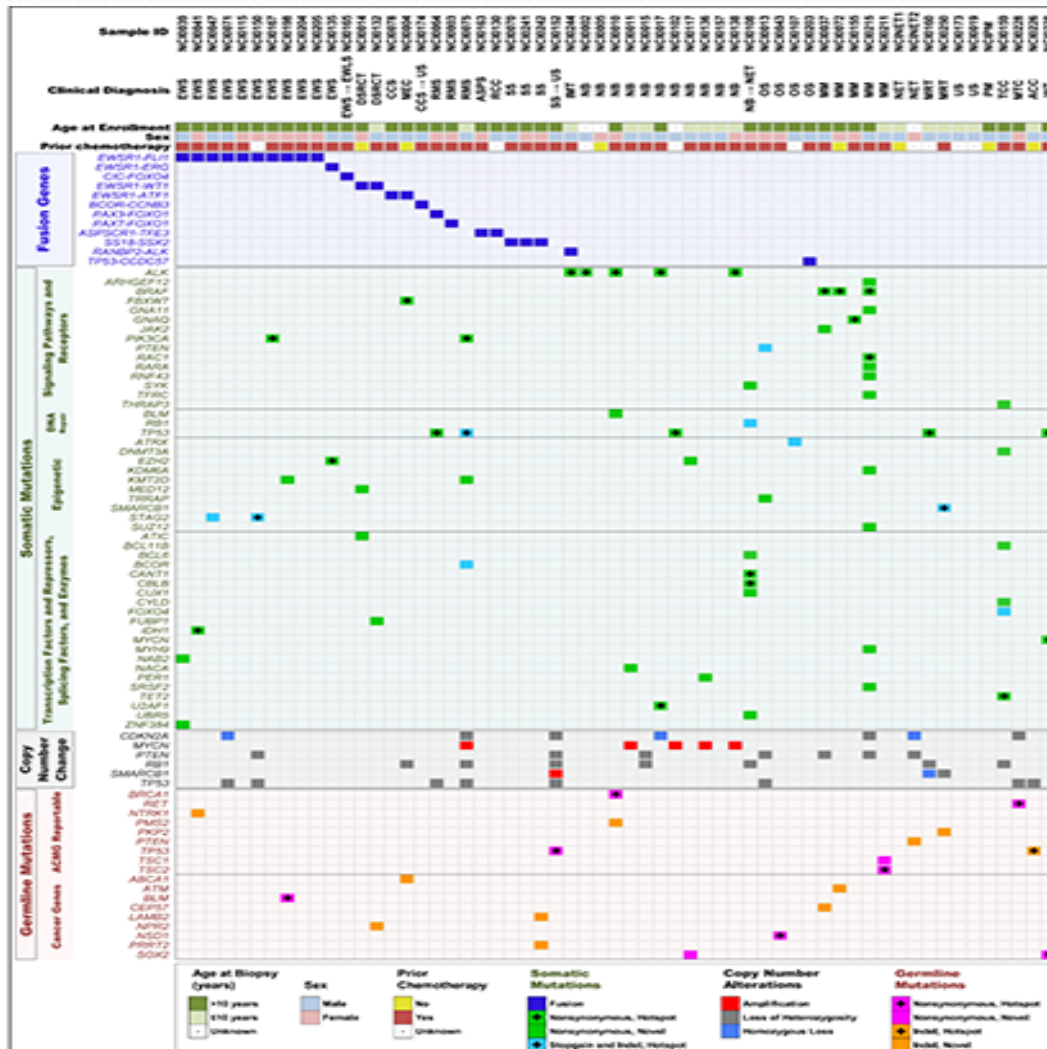
Title: “Comprehensive Omics Analysis of Pediatric Solid Tumors and Establishment of a Repository for Related Biological Studies” or Omics protocol

Study design

Study Design

- Pilot study to determine the utility and feasibility of performing comprehensive genomic analyses to identify clinically actionable mutations in pediatric and young adult patients with metastatic, refractory or relapsed solid tumors
- 59 patients enrolled to the pediatric oncology branch, Center for Cancer Research (CCR), NCI (2010-2014)
- Age 7 months-25 years
- 20 diagnostic categories (non-CNS, solid tumors)
- Comprehensive multi-omics exome germline & tumor, RNAseq tumor & Illumina Omni SNP arrays of tumor

Multi-omics integrated landscape



Multi-Omics Integrated Landscape

RNAseq
Diagnostic, Driver, Actionable

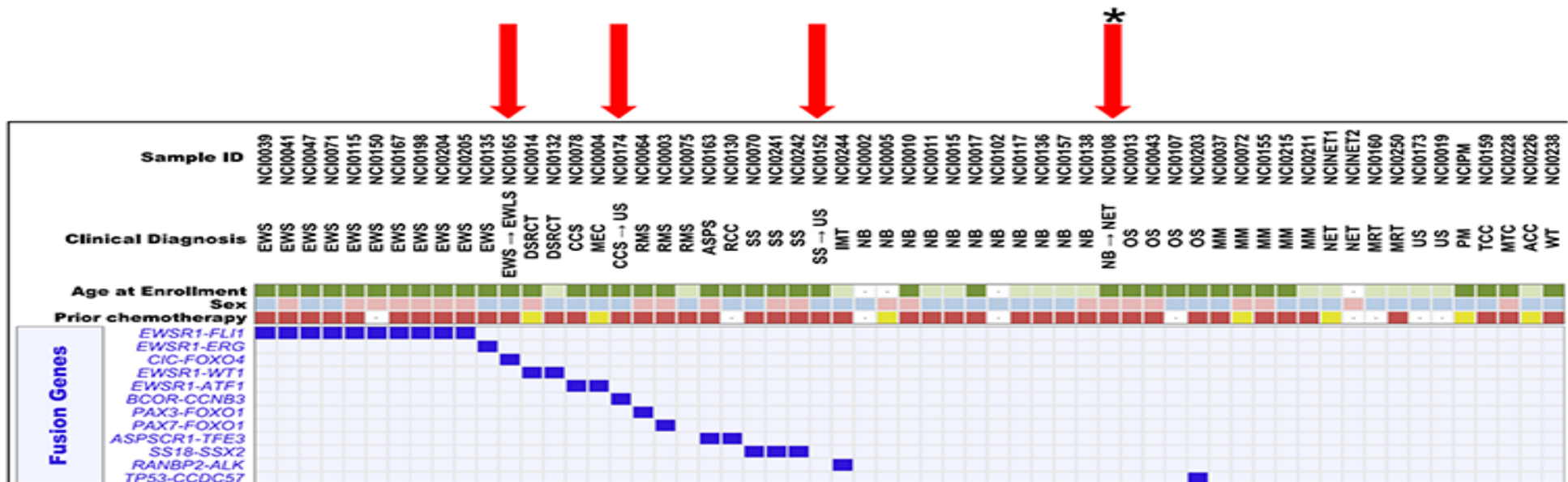
DNAseq and RNAseq
Somatic: Driver, Actionable

DNA copy number & RNAseq
Somatic: Driver, Actionable

DNAseq
Germ line: Disease causing, Actionable



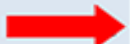

Fusion genes

Presence or absence of fusion genes and/or expression profiles confirms diagnosis or leads to revision of diagnosis



Germline mutations

~10% of Pediatric and Adolescent Young Adults with Cancers have Actionable Germline Mutations some Therapeutically

Sample	Diagnosis	Gene	Mutation	Disease	Hotspot	Notes	ACMG gene
NCI0072	MM	<i>ATM</i>	p.Y380fs	Ataxia-Telangiectasia and Cancer Predisposition Syndrome	No	Frameshift Insertion of Tumor Suppressor Gene	Yes
NCI0010	NB	<i>BRCA1</i>	Q1313X	Hereditary Breast and Ovarian Cancer Syndrome	Yes	Pathogenic, Reportable	Yes
NCI0010	NB	<i>PMS2</i>	p.K356fs	Lynch Syndrome and Mismatch Repair Cancer Syndrome	No	Frameshift Deletion of Tumor Suppressor Gene	Yes
	NET	<i>PTEN</i>	p.R14fs	PTEN Hamartoma Tumor Syndrome	No	Frameshift Deletion of Tumor Suppressor Gene	Yes
	MTC	<i>RET</i>	M918T	Multiple Endocrine Neoplasia 2B	Yes	Pathogenic, Reportable	Yes
NCI0152	SS → US	<i>TP53</i>	R175H	Li-Fraumeni Syndrome	Yes	Patient Tumor has LOH of Wild-Type TP53 on Other Allele	No
NCI0226	ACC	<i>TP53</i>	A159K	Li-Fraumeni Syndrome	Yes	Tumor has LOH of Wild-Type TP53 on Other Allele, Novel, 2 Base Non-Frameshift Substitution, c.358_359delGCinsTT	No
	MM	<i>TSC1</i>	p.S828R	Tuberous Sclerosis Type 1, Lymphangioleiomyomatosis, Focal Cortical Dysplasia, and Everolimus Sensitivity	No	Nonsynonymous SNV, Autosomal Dominant, Patient also has a Germline TSC2 Mutation	No
	MM	<i>TSC2</i>	p.T246A	Tuberous Sclerosis Type 2, and Lymphangioleiomyomatosis	Yes	Nonsynonymous SNV, Autosomal Dominant, Patient also has a Germline TSC1 Mutation	No

Tumor mutations

Approximately 50% of Pediatric and Adolescent Young Adults with Cancers have Actionable Tumor Mutations

Sample	Diagnosis	Gene	Stage	Modality	AA Change	Level	Drug	Clinical Trial: Pediatric	FDA Approval in Adults	Exact Mutation vs. Hotspot
NCI0041	EWS	IDH1	Relapsed	WES/WT	p.R132C	2a	IDH1 inhibitors	No	No	Exact
NCI0167	EWS	PK3CA	Refractory	WES/WT	p.D1017G	2a	PI3K/AKT/mTOR inhibitors	Yes	Yes	Exact
NCI0071	EWS	CDKN2A	Relapsed	SNP Array/WT	Homozygous loss	3	CDK4/6 inhibitor	No	No	-
NCI0047	EWS	STAG2	Relapsed	WES/WT	p.E984X	3	PARP inhibitors	Yes	No	-
NCI0150	EWS	STAG2	-	WES/WT	p.R216X	3	PARP inhibitors	Yes	No	Hotspot
NCI0244	IMT	ALK	Relapsed	WT	RANBP2-ALK fusion	2a	Crizotinib	No	Yes	Exact
NCI0244	IMT	ALK	Relapsed	WES/WT	p.I1171T	2a	Crizotinib	No	Yes	Exact
NCI0037	MM	BRAF	Relapsed	WES/WT	p.V600E	1	Vemurafenib, Dabrafenib	Yes	Yes	Exact
NCI0072	MM	BRAF	Diagnostic	WES/WT	p.V600E	1	Vemurafenib, Dabrafenib	Yes	Yes	Exact
NCI0215	MM	BRAF	Relapsed	WES/WT	p.V600E	1	Vemurafenib, Dabrafenib	Yes	Yes	Exact
NCI0155	MM	GNAQ	Relapsed	WES/WT	p.Q209L	1	Tamoxifen, Trametinib, Vorinostat	No	Yes	Exact
NCI0215	MM	GNA11	Relapsed	WES/WT	p.S268F	2a	Trametinib	No	Yes	-
NCI0211	MM	TSC1	Relapsed	WES/WT	p.S828R	3	Everolimus	No	Yes	-
NCI0211	MM	TSC2	Relapsed	WES/WT	p.T246A	3	Everolimus	No	Yes	-
NCI0160	MRT	SMARCB1	-	SNP Array/WT	Homozygous loss	3	EZH2 inhibitors	No	No	-
NCI0250	MRT	SMARCB1	Refractory	WES/WT	p.R40X	3	EZH2 inhibitors	No	No	-
NCI0228	MTC	RET	Relapsed	WES/WT	p.M918T	2a	Vandetanib	Yes	Yes	Exact
NCI0002	NB	ALK	-	WES/WT	p.R1275Q	2a	Crizotinib	Yes	Yes	Exact
NCI0010	NB	ALK	Relapsed	WES/WT	p.F1174V	2a	Crizotinib	Yes	Yes	Exact
NCI0017	NB	ALK	Relapsed	WES/WT	p.F1174L	2a	Crizotinib	Yes	Yes	Exact
NCI0138	NB	ALK	Relapsed	WES/WT	p.Y1278S	2a	Crizotinib	Yes	Yes	Exact
NCI0017	NB	CDKN2A	Relapsed	SNP Array/WT	Homozygous loss	3	CDK4/6 inhibitor	No	No	-

Sample	Diagnosis	Gene	Stage	Modality	AA Change	Level	Drug	Clinical Trial: Pediatric	FDA Approval in Adults	Exact Mutation vs. Hotspot
NCI0011	NB	MYCN	Relapsed	SNP Array/WT	Amplification	3	bromodomain inhibitors	No	No	-
NCI0102	NB	MYCN	-	SNP Array/WT	Amplification	3	bromodomain inhibitors	No	No	-
NCI0136	NB	MYCN	Relapsed	SNP Array/WT	Amplification	3	bromodomain inhibitors	No	No	-
NCI0138	NB	MYCN	Relapsed	SNP Array/WT	Amplification	3	bromodomain inhibitors	No	No	-
NCINET2	NET	PTEN	-	WES/WT	p.R14fs	2a	PI3K/AKT/mTOR inhibitors	Yes	No	-
NCINET2	NET	CDKN2A	-	SNP Array/WT	Homozygous loss	3	CDK4/6 inhibitor	No	No	-
NCI0013	OS	PTEN	Relapsed	WES/WT	p.K80fs	2a	PI3K/AKT/mTOR inhibitors	Yes	No	-
NCI0075	RMS	PK3CA	Relapsed	WES/WT	p.P104Q	2a	PI3K/AKT/mTOR inhibitors	Yes	Yes	Exact
NCI0075	RMS	MYCN	Relapsed	SNP Array/WT	Amplification	3	bromodomain inhibitors	No	No	-
NCI0238	WT	MYCN	Relapsed	WES/WT	p.P44L	3	bromodomain inhibitors	No	No	-

NCI-Adult MATCH Criteria for Matching Mutation to Drug

Level 1	Gene variant approved for selection of an approved drug (BRAF V600E and vemurafenib). The variant will be Level 1 in all tissues open to treatment with the approved drug.
Level 2a	Gene variant is an eligibility criteria for an ongoing clinical trial for that treatment.
Level 2b	Gene variant has been identified in an N of 1 responses (TSC1 and everolimus) for that treatment.
Level 3	Preclinical inferential data (in vivo and in vitro models) that provide biological evidence sufficient to support the use of a variant for treatment selection, e.g. <ul style="list-style-type: none"> Models with variants respond to treatment and models without variant do not respond to treatment Gain of function mutations demonstrated in pre-clinical model, e.g. D769H variant of ERBB2 results in increased tyrosine kinase-specific activity and up regulates pathway signaling (does not require treatment evidence) Loss of function genes, tumor suppressor or pathway inhibitor (e.g. NF1) any variant that produces a stop codon including frameshift or demonstrated loss of function in pre-clinical model (does not require treatment evidence)

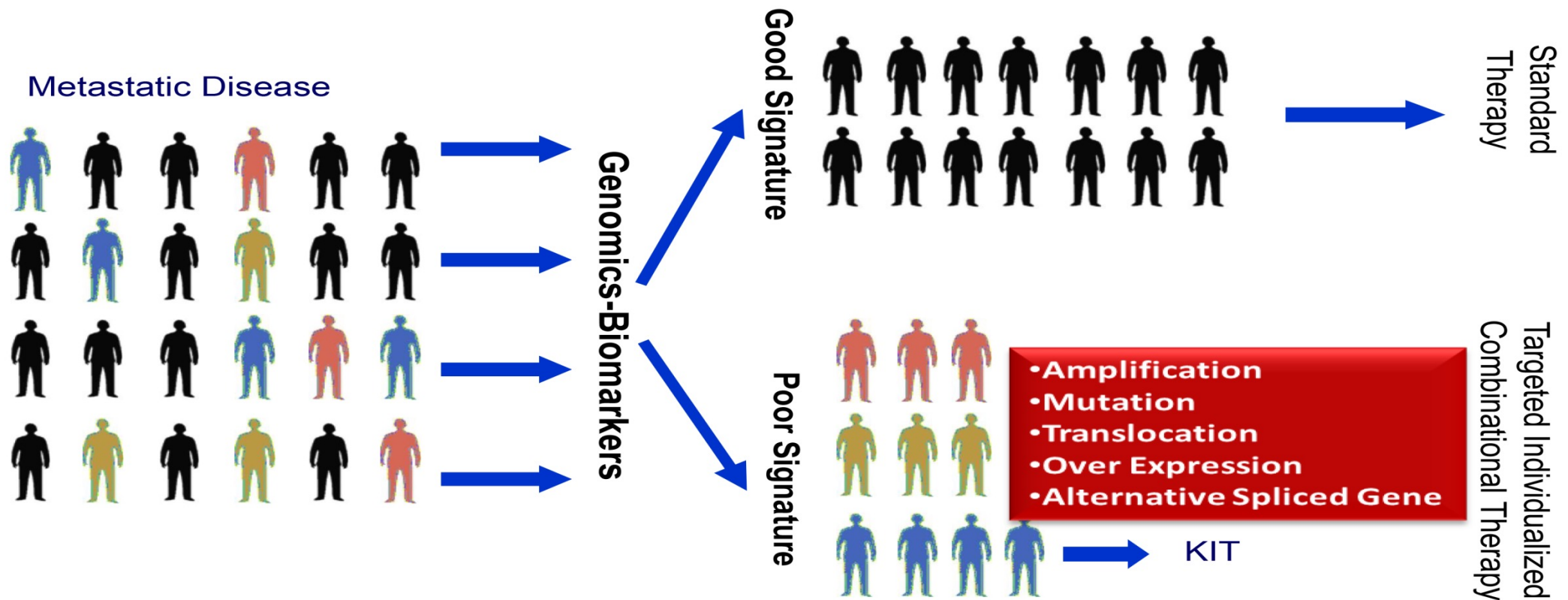
Summary

Summary

- Demonstrated the importance and feasibility of performing multi-dimensional ClinOmics in the clinical setting in real time
- ~50% of children with pediatric or AYA patients with relapsed or refractory cancers have actionable somatic mutations
- ~ 10% have actionable germline mutations
- Importance of performing parallel germline sequencing; some therapeutically actionable (e.g. DNA repair, PTEN, TSC1, TSC2, HRAS, RET, ALK)
- Increased tumor burden in relapsed tumors; implications for immunotherapy
- Single agent pediatric MATCH like trials are planned by COG-NCI

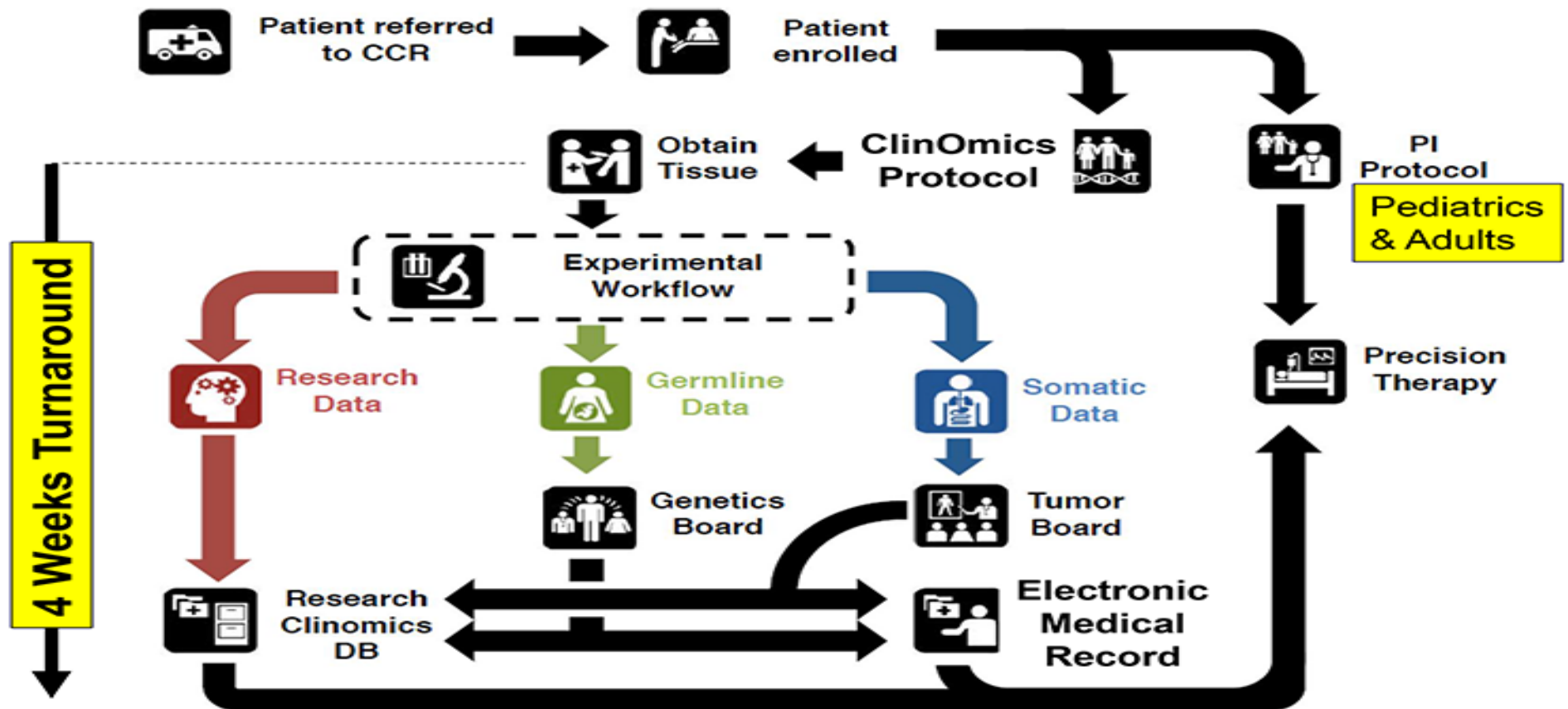
Future Trials

Genomics Enabling Precision Therapy-The Future for Pediatric Trials



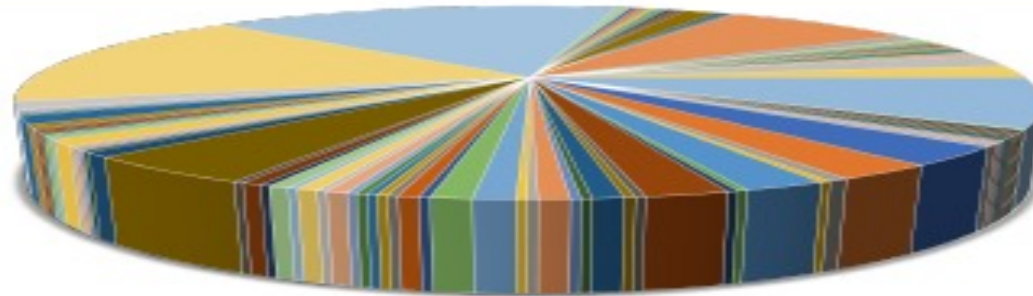
ClinOmics program

CCR ClinOmics Program-CLIA



Patient diagnoses

396 Patients of 93 diagnoses



BRAC

- (1) Anaplastic Astrocytoma
- (2) Anaplastic Oligo
- (3) Bladder cancer
- (4) Cholangiocarcinoma
- (5) Dermatofibrosarcoma protuberans
- (6) Diffuse intrinsic pontine glioma
- (7) Ependymoma
- (8) Glioblastoma
- (9) Glioblastoma
- (10) Grade II Oligodendroglioma
- (11) Invasive well differentiated squamous cell carcinoma
- (12) Lymphocytoma
- (13) Melanoma
- (14) Mesothelioma Pleural
- (15) Metastatic Pancreatic Neuroendocrine Carcinoma
- (16) Multiple Retic Tumors
- (17) Neuroblastoma I
- (18) Osteosarcoma
- (19) Papillary tumor of the pineal region
- (20) Poorly differentiated carcinoma (lung vs. thyroid)
- (21) Renal cell carcinoma
- (22) Small Cell Cancer of rectum
- (23) Temporal high grade glioma
- (24) Uveal melanoma

- (25) Acute lymphoblastic leukemia
- (26) Anaplastic Ependymoma
- (27) Anaplastic Fibrous Histiocytoma
- (28) Breast cancer
- (29) Chondroma
- (30) Desmoid Fibromatosis
- (31) Endometrial cancer
- (32) Ewing's sarcoma
- (33) Glioblastoma
- (34) Hepatic Angiosarcoma
- (35) Keratoacanthoma
- (36) Melanocytoma
- (37) Merkel Cell Carcinoma
- (38) Mesothelioma Thoracic
- (39) MPPST
- (40) Myxopapillary Ependymoma
- (41) Neuronal tumor
- (42) Ovarian Serous Carcinoma
- (43) Piloepithelioma
- (44) Prostate cancer
- (45) Pseudopapilloma
- (46) Small cell carcinoma of the ovary hypercalcemic type (OCEH)
- (47) Teratoma

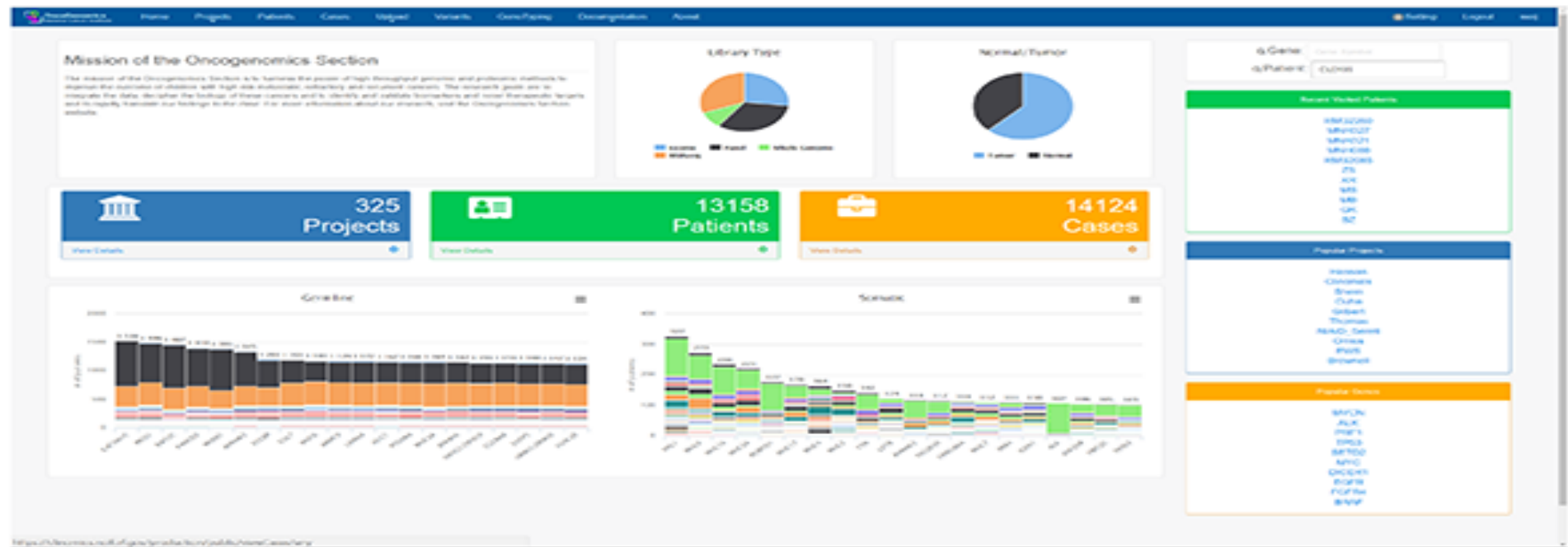
- (48) Acute myeloid leukemia
- (49) Anaplastic meningioma
- (50) Astrocytoma
- (51) Carcinoid, BRCA1 positive
- (52) Clear cell sarcoma
- (53) Desmoplastic small round cell tumor
- (54) Endometrial Stromal Sarcoma
- (55) Extracranial small cell cancer
- (56) Glioma
- (57) Hepatocellular cancer
- (58) Left Convoluted Sarcoma
- (59) Medullary Thyroid Cancer metastatic
- (60) Mesothelioma
- (61) Metastatic Adip Carcinoma
- (62) Multinodular and Vasculating Neuronal Tumor
- (63) Neuroendocrine carcinoma
- (64) Neuroendocrine giant cell tumor
- (65) Ovarian Teratoma
- (66) Piloepithelioma
- (67) Pleomorphic Xanthoastrocytoma
- (68) Recurrent gliosarcoma tumor
- (69) SDC
- (70) Small cell endometrium
- (71) Thyroid

- (72) Ampullary cancer
- (73) Anaplastic Oligodendroglioma
- (74) Atypical Central Neurocytoma
- (75) Carcinoma of the Pelvis
- (76) Colon cancer
- (77) Diffuse Astrocytoma, Grade II
- (78) Endophylla
- (79) Gallbladder cancer
- (80) Gliosarcoma
- (81) Hepatocellular carcinoma
- (82) Lung Adenocarcinoma
- (83) Medulloblastoma
- (84) Mesothelioma Peritoneal
- (85) Metastatic NET
- (86) Multiple carcinoma
- (87) Neuroendocrine Tumor
- (88) NGLC
- (89) Pancreatic cancer
- (90) Pleomorphic xanthoastrocytoma
- (91) Recurrent Medulloblastoma
- (92) Small cell bladder
- (93) Synovial sarcoma
- (94) Undifferentiated sarcoma

ClinOmics Data Portal

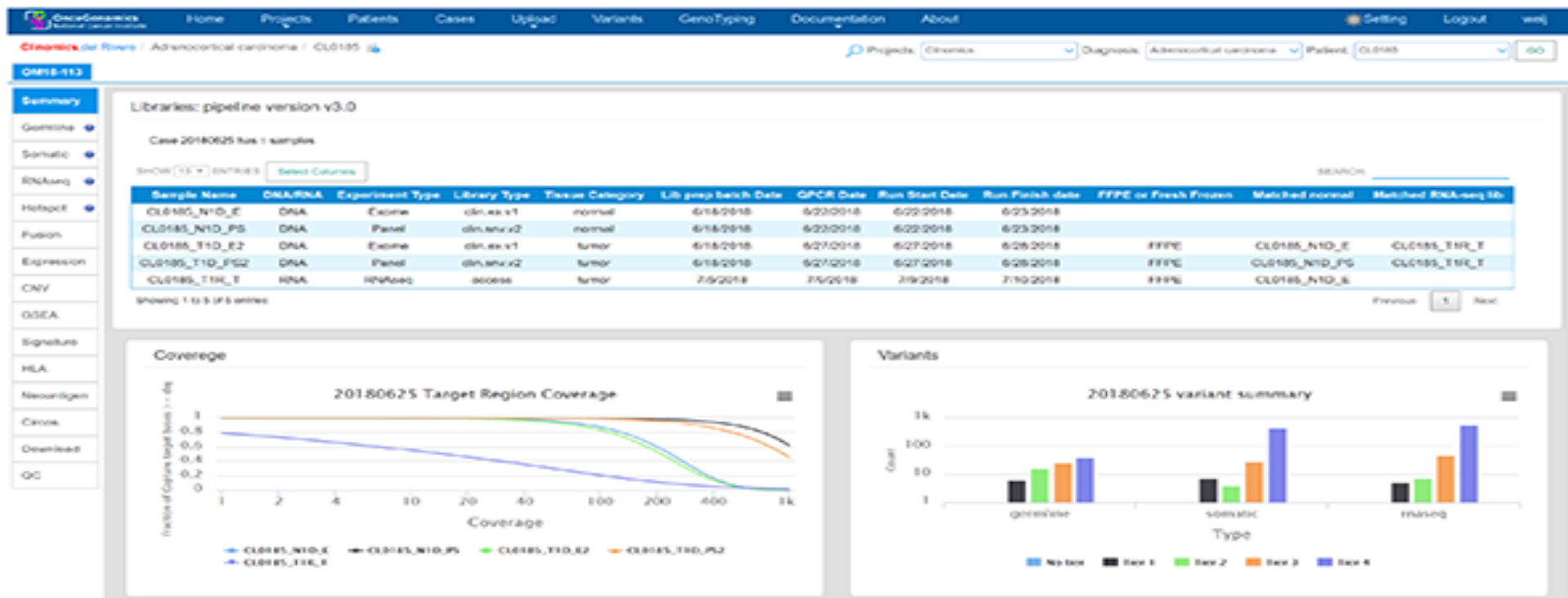
ClinOmics Data Portal

<https://clinomics.ncifcrf.gov/production/public/>



Patient Summary

Patient Summary Page



QC report

QC Report: Sequencing Statistics & Genotyping

Run Statistics

Summary | Clones | Coverage | Transcript Coverage | Hotspot | Contours | Compare | **DNA QC** | RNA QC | RNA QC v2 | FASTQC | Genotyping | Versions

Genome Solid colour

Search:

Sample_ID	MEAN PAIR COVERAGE	MEAN TARGET COVERAGE	Total reads	Mapped reads	Percent mapped	On-target reads	Percent on-target	Unique on-target reads	Percent unique on-target	Hq unique on-target reads	Percent Hq unique on-target	Percent Hq unique positions at 20x	Percent Hq unique positions at 30x	Percent Hq unique positions at 50x	Percent Hq unique positions at 100x	Percent Hq unique positions at 200x	Percent Hq unique positions at 400x
CL0185_N1D_E_H2WNCBGK7	190	210	250645548	257511346	98.55	168950939	95.96	137432996	86.81	132687067	86.55	96.41	95.53	92.74	79.74	43.42	5.53
CL0185_N1D_PS_H2WNCBGK7	888	781	61489380	61218679	98.80	41885021	68.43	26281444	62.86	25877230	97.81	98.38	98.30	98.16	97.75	96.80	96.75
CL0185_T1D_E2_HLJYOBGK7	170	182	230619044	237360954	98.48	153908821	64.67	124326705	86.74	120344211	96.72	96.06	94.78	90.70	74.07	35.34	3.91
CL0185_T1D_PS2_HLJYOBGK7	876	833	56426879	60183182	98.88	34227908	61.84	21781328	63.35	21288182	97.78	98.33	98.25	98.04	96.88	91.71	72.70

Showing 1 to 4 of 4 entries

Genotyping

Summary | Clones | Coverage | Transcript Coverage | Hotspot | Contours | Compare | DNA QC | RNA QC | RNA QC v2 | FASTQC | **Genotyping** | Versions

Genome Solid colour

Comment...

☒ Pass ☐ Fail

Search:

Sample	CL0185_N1D_E	CL0185_N1D_PS	CL0185_T1D_E2	CL0185_T1D_PS2	CL0185_T1D_T
CL0185_N1D_E	100%	100%	100%	99%	99%
CL0185_N1D_PS	100%	100%	100%	99%	99%
CL0185_T1D_E2	100%	100%	100%	99%	99%

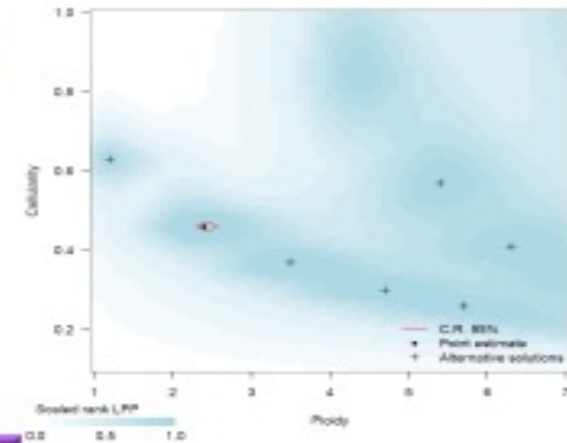
QC Report: Coverage

QC Report: Coverage

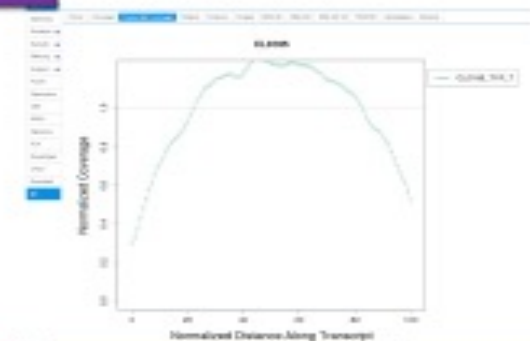
Circos



Tumor Content



RNA Coverage



Hotspot Coverage



Germline and Somatic Mutations

[illegible]

EGFR mutations

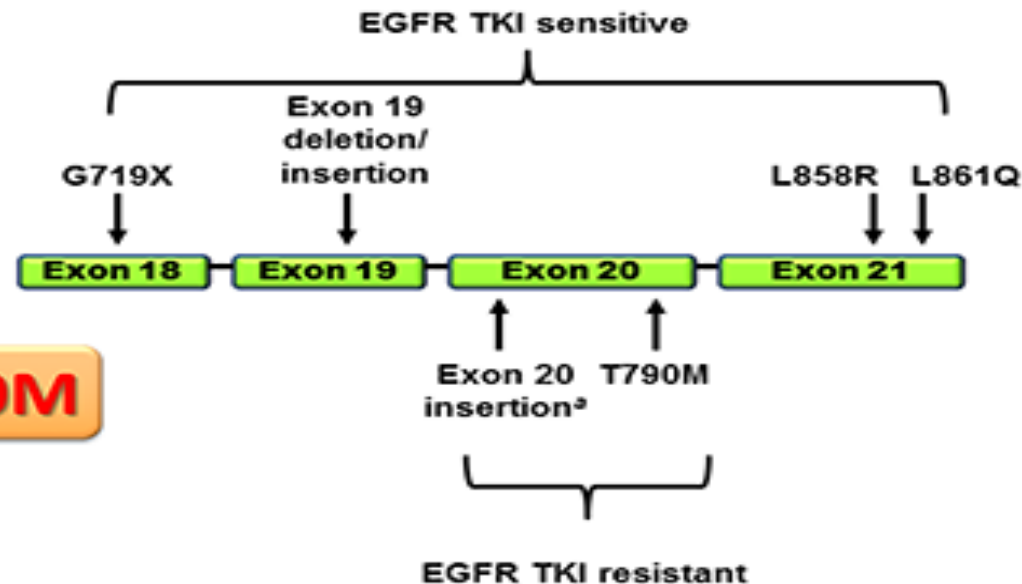
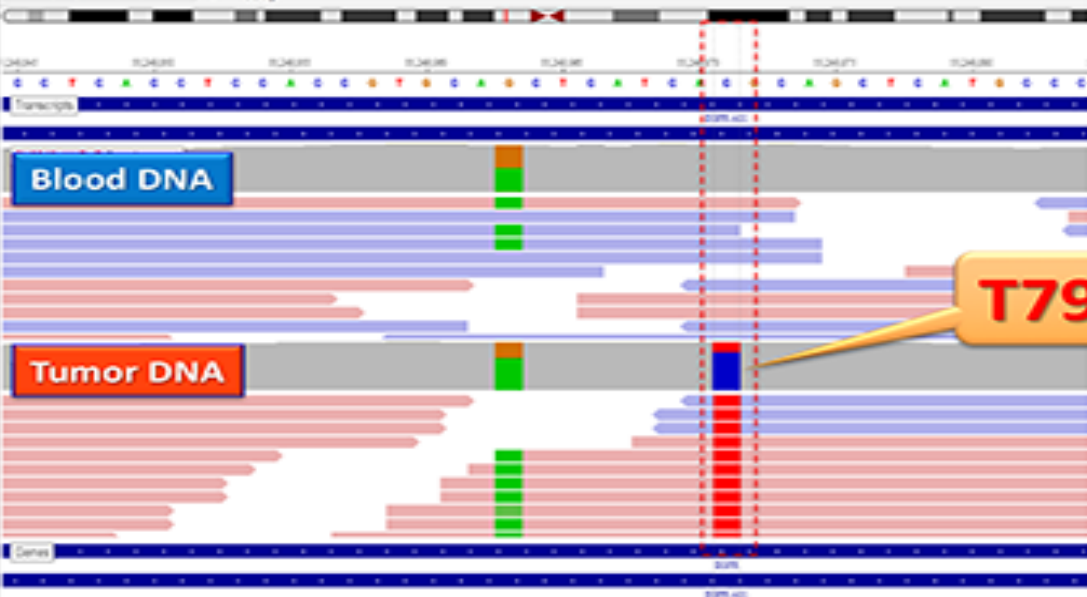
EGFR mutations in NSCLC

GV view of patient: CL0040 case: OM16-007 Total 4 sample(s)

ic: (check sample to load)

CL0040_T1D_E Exome, normal (Exome, normal) CL0040_T1D_E Exome, tumor (Exome, tumor) CL0040_T1D_P Panel, normal (Panel, normal) CL0040_T1D_P Panel, tumor (Panel, tumor)

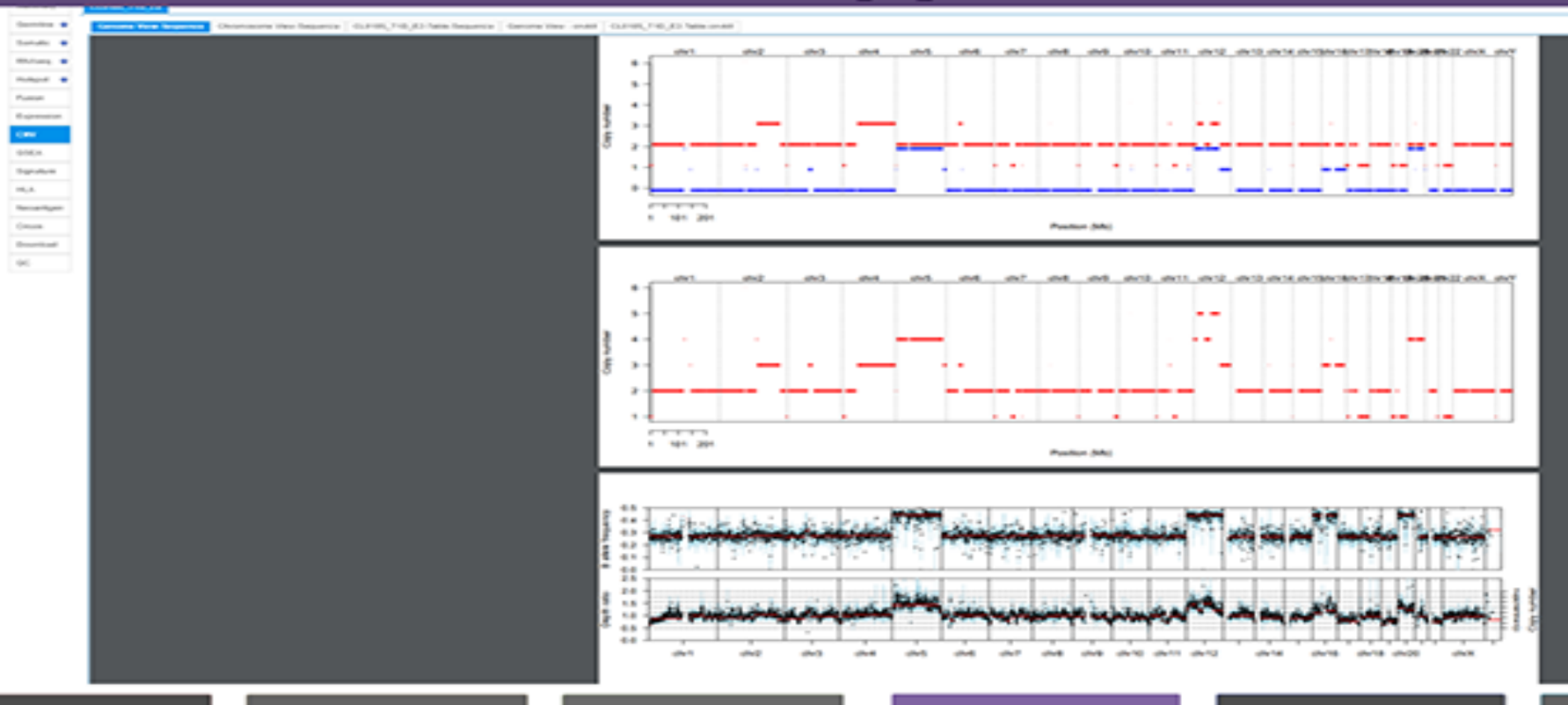
chr7:55,243,244-55,249,267 53 bp



<https://www.mycancergenome.org/content/disease/lung-cancer/egfr/>

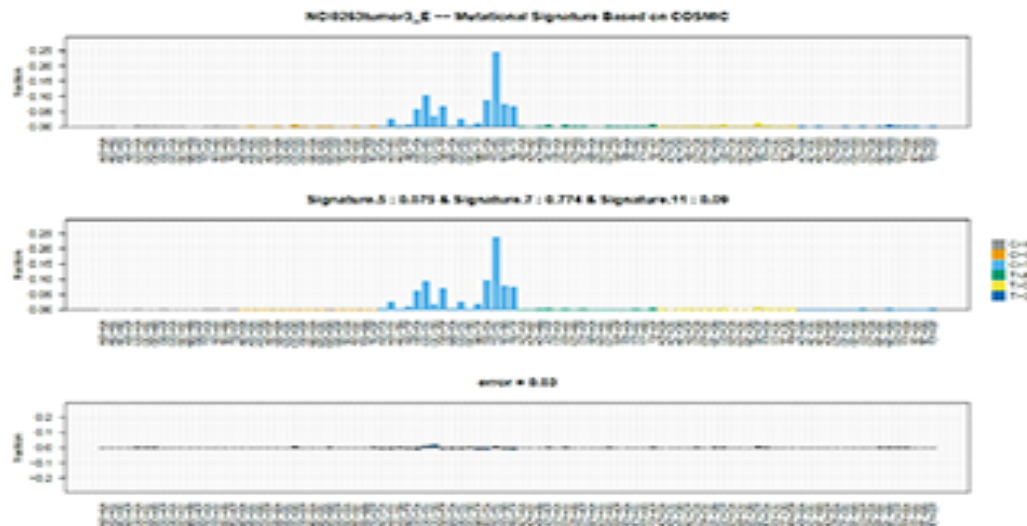
Tumor Copy Number

Tumor Copy Number

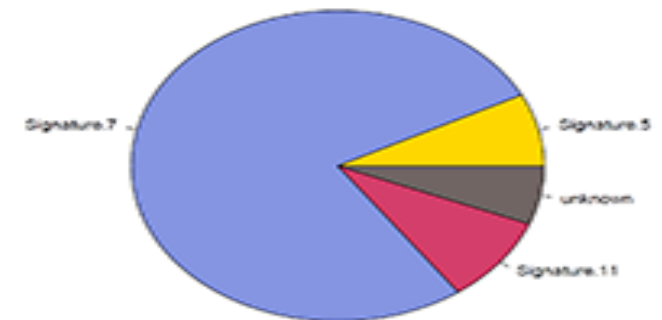


Mutation Signatures

Mutation Signatures for Tumor

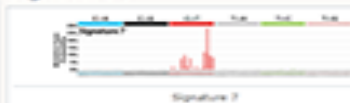


NCI0263: Melanoma



COSMIC (<https://cancer.sanger.ac.uk/cosmic/signatures>)

Signature 7



Cancer types: Signature 7 has been found predominantly in skin cancers and in cancers of the lip categorized as head and neck or oral squamous cancers.

Proposed aetiology: Based on its prevalence in ultraviolet exposed areas and the similarity of the mutational pattern to that observed in experimental systems exposed to ultraviolet light Signature 7 is likely due to ultraviolet light exposure.

Additional mutational features: Signature 7 is associated with large numbers of CC>TT dinucleotide mutations at dipyrimidines. Additionally, Signature 7 exhibits a strong transcriptional strand bias indicating that mutations occur at pyrimidines (via, e.g. formation of pyrimidine-pyrimidine photoproducts) and these mutations are being repaired by transcription-coupled nucleotide excision repair.

Cosmicdb: N/A

Signature 7: UV signature

Mutation Burden

Mutation Burden

The screenshot displays the OncoGenomics National Cancer Institute web application. The top navigation bar includes links for Home, Projects, Patients, Cases, Upload, Variants, GenoTyping, Documentation, and About. A user profile 'weij' is logged in. The breadcrumb trail shows 'Clinomics, del Rıvero / Adrenocortical carcinoma / CL0185'. Below this, filters for Projects (Clinomics), Diagnosis (Adrenocortical carcinoma), and Patient (CL0185) are set, with a 'GO' button. The left sidebar lists various analysis types: Summary, Germline, Somatic (selected), RNAseq, Hotspot, Fusion, Expression, CNV, GSEA, Signature, and HLA. The main content area is titled 'OM18-113' and shows tabs for 'Somatic-All', 'Somatic-CL0185_T1D_PS2-Panel', 'Somatic-CL0185_T1D_E2-Exome', and 'Mutation_Burden' (selected). Under 'Mutation_Burden', the 'Callers' dropdown is set to 'MuTect'. A 'Select Columns' button is visible. The table shows 2 records out of 6 total entries, filtered from 6 total entries. The table has columns: Diagnosis, Sample Name, Experiment Type, Caller, Burden, Total bases, and Burden Per MB. The 'Burden Per MB' column is highlighted with a red box. The table data is as follows:

Diagnosis	Sample Name	Experiment Type	Caller	Burden	Total bases	Burden Per MB
Adrenocortical carcinoma	CL0185_T1D_E2	Exome	MuTect	612	45196537	13.54
Adrenocortical carcinoma	CL0185_T1D_PS2	Panel	MuTect	36	2465827	14.6

Showing 1 to 2 of 2 entries (filtered from 6 total entries)

Navigation: Previous 1 Next

Fusion Gene Detection

Fusion Gene Detection from RNA-seq experiments



Useful Genomic Information

Other Useful Genomic Information

- **HLA typing (Tissue typing)**
- **Neoantigen prediction**
- **Gene expression**
- **Gene Set Enrichment Analysis (GSEA)**
- **Survival analysis if outcome data is available**

Conclusions:

Next generation sequencing (including whole genome, exome and transcriptome) determines the complete genomic and epigenetic portrait of cancers at the base pair level.

Integrated analyses of the cancer can identify biologically relevant diagnostic, prognostic biomarkers and novel targets for precision medicine.

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