

Genomics

Apply Genomics to Precision Medicine

*Jun S. Wei, Ph.D.
Oncogenomics Section
Genetics Branch
Center for Cancer Research
National Cancer Institute*

*TRACO
September 14, 2026*

Outline

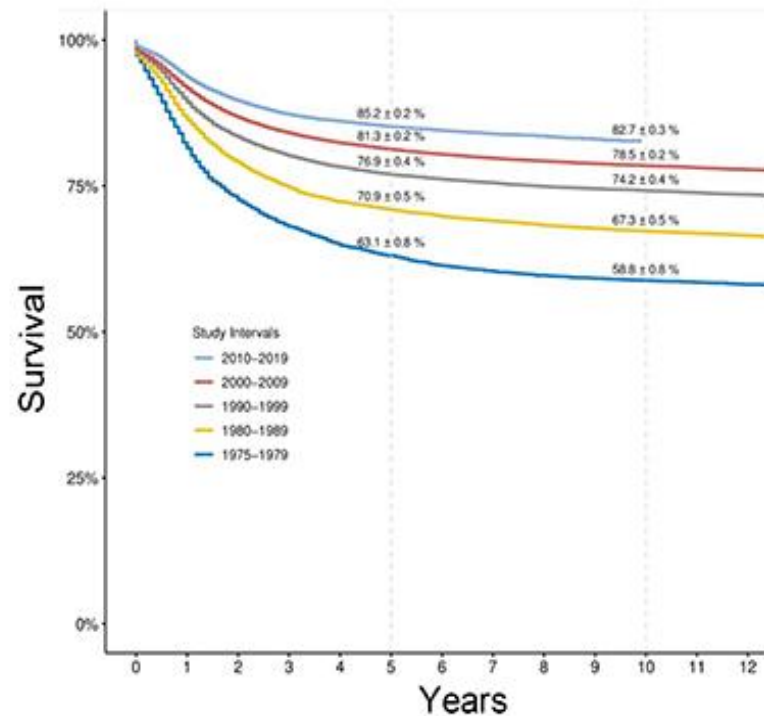
Outline

- **Success and Challenges of Treating Pediatric Cancers**
- **Genomics**
- **Tool to study genomics: Next-generation Sequencing**
- **Precision medicine – an application of genomics**

Childhood cancer

National Cancer Institute

Childhood cancer: The beginning of a modern medical success story



Sultan et. al., PloS One 2025. PMID: 39752445

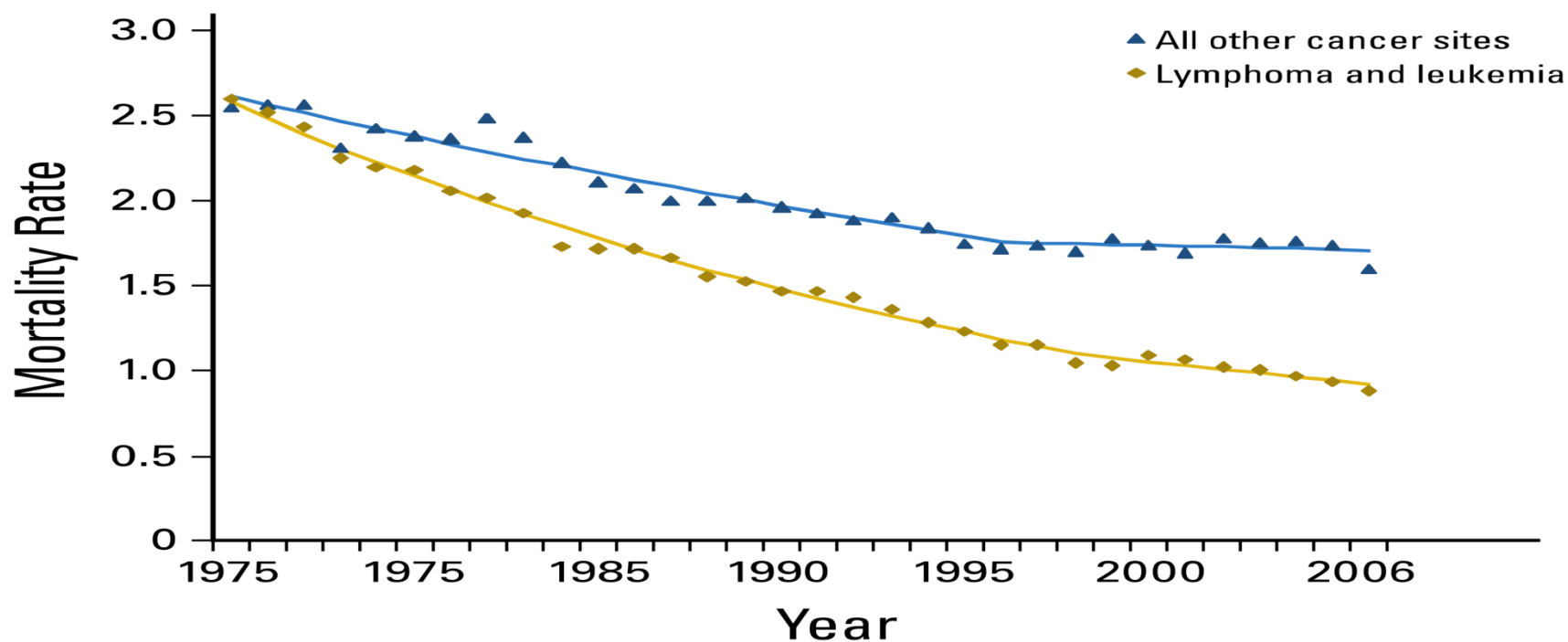


Courtesy: John Maris

Mortality rates

National Cancer Institute

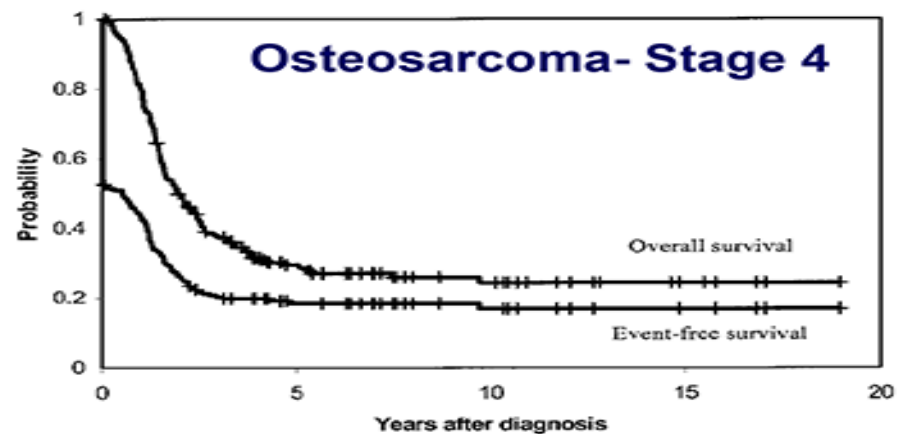
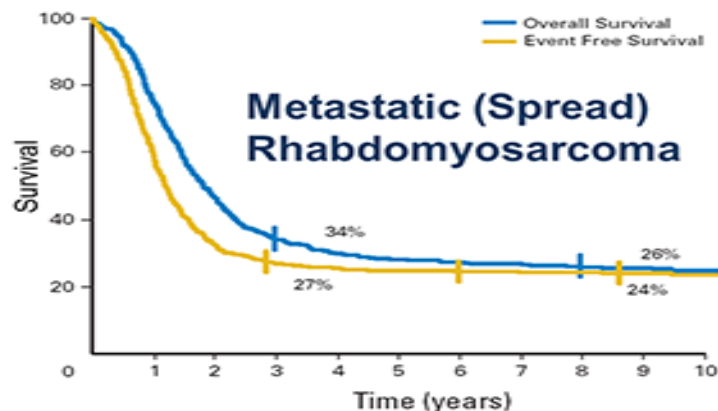
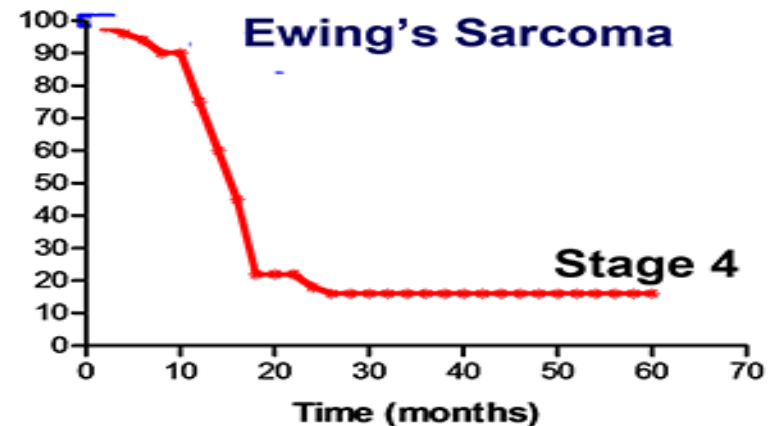
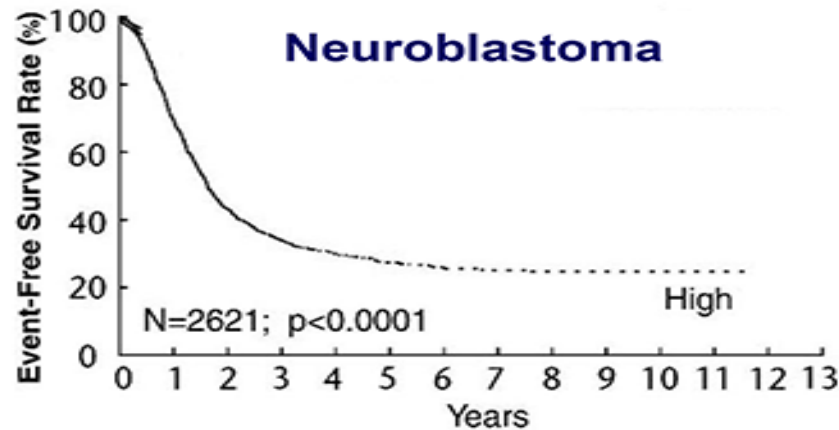
However in the past 16 years no improvement in mortality rates despite increased intensity of treatment



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

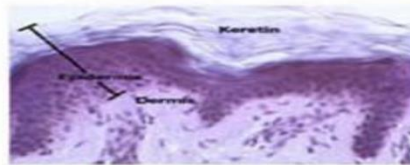


Gene expression

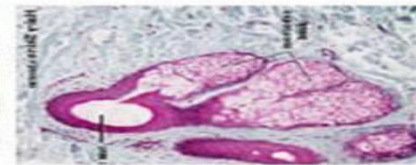
...but the complexity and diversity

Same genome or DNA →

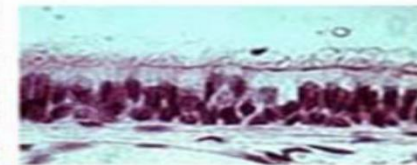
- Different expression pattern
- Different proteome
- Different tissues
- Different physiology



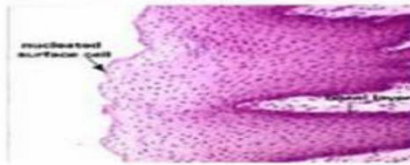
skin



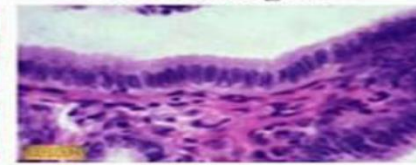
sebaceous gland



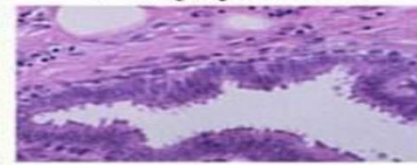
airway epithelium



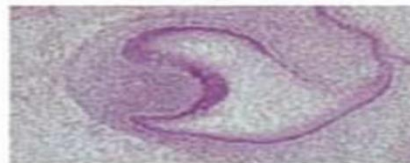
tongue



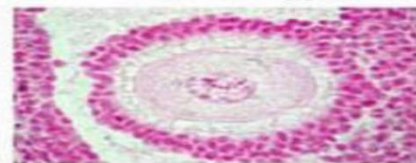
intestinal crypt



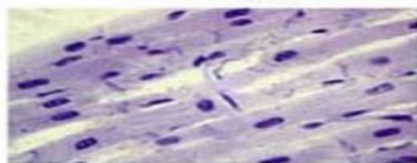
mammary gland



developing tooth



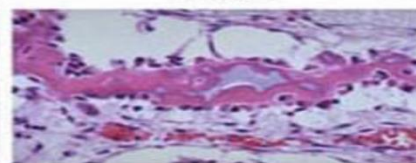
follicle



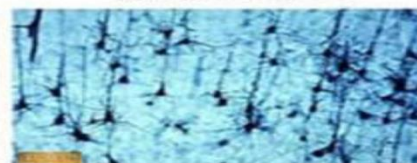
skeletal muscle



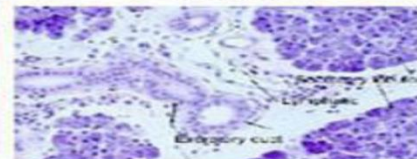
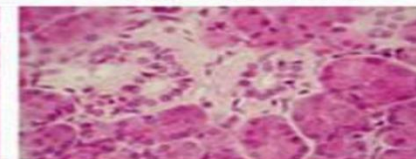
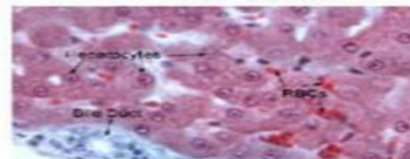
developing bone



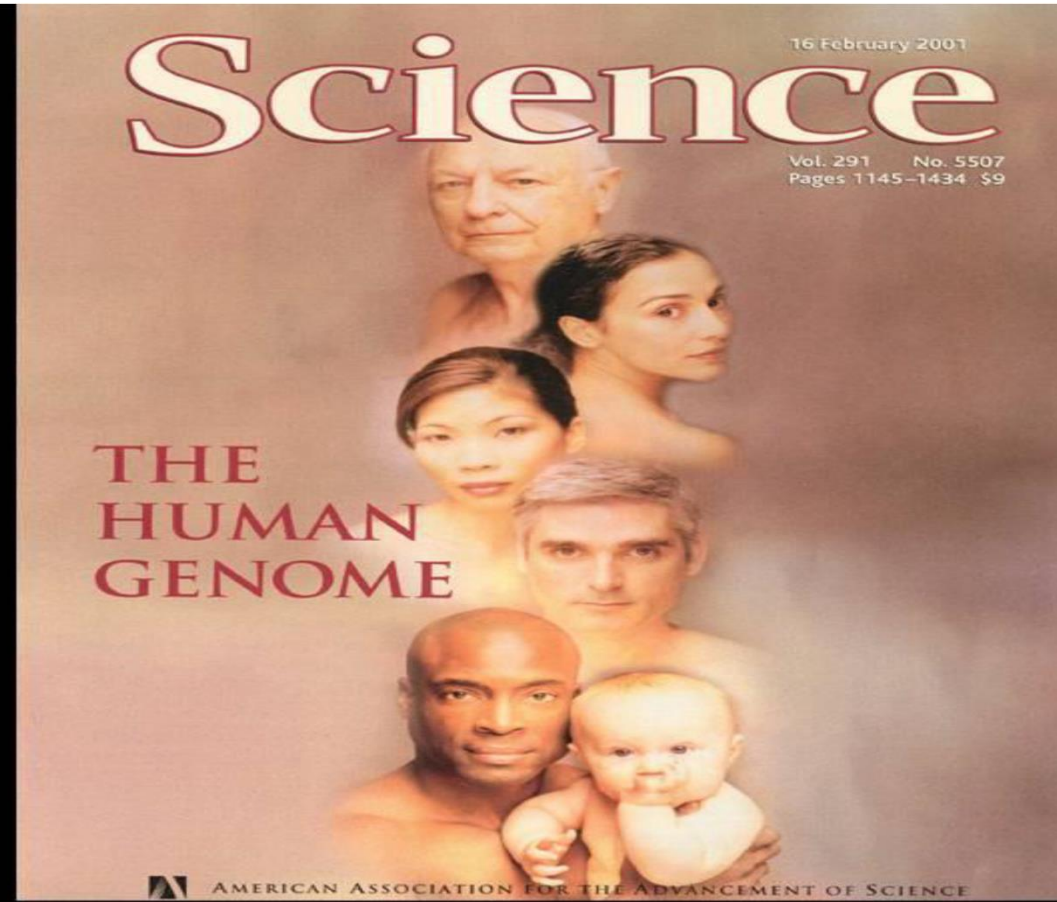
bone (high mag)



neuron

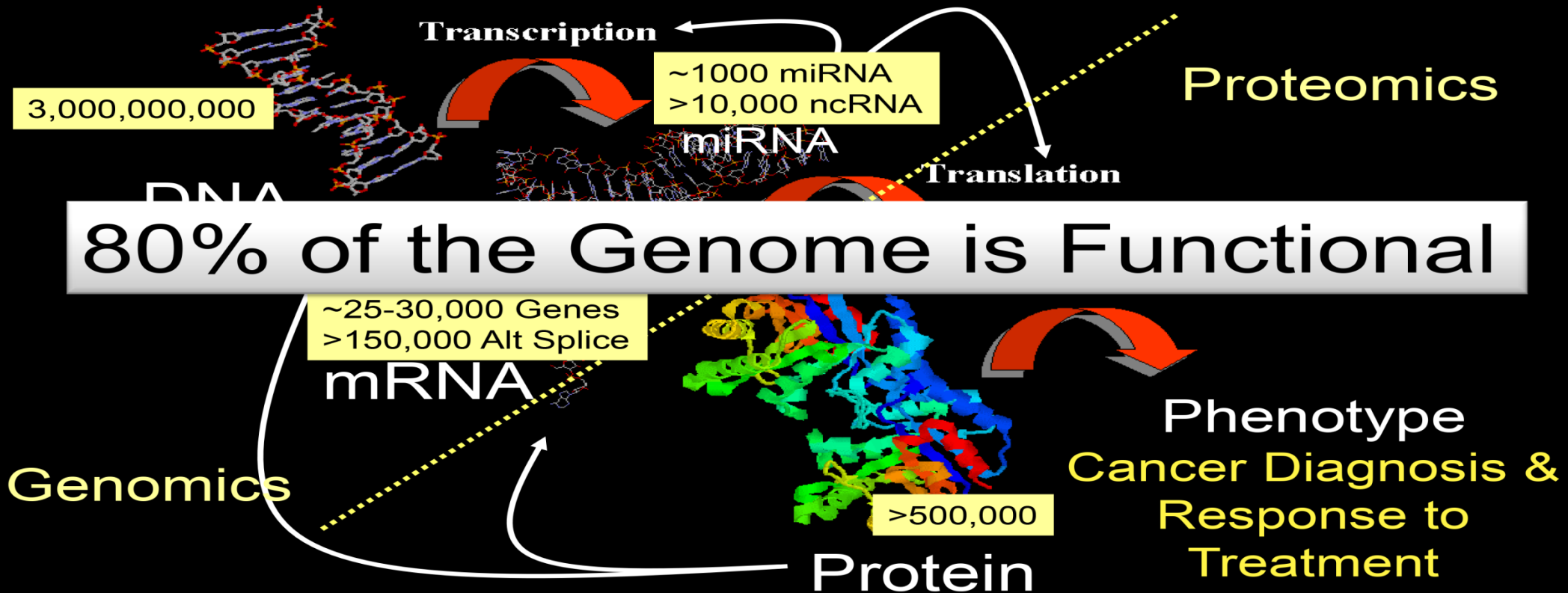


Human genome



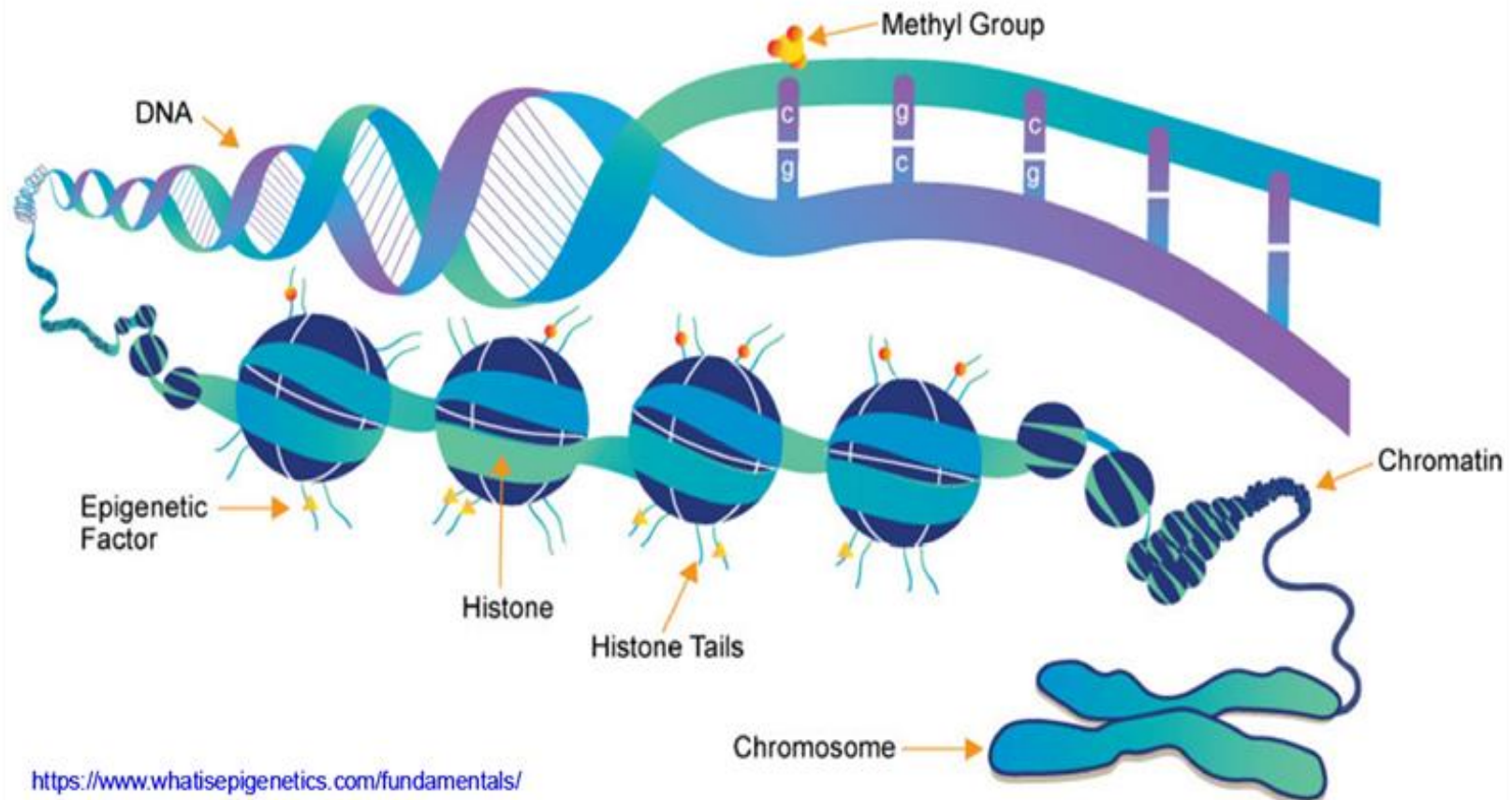
Gene expression

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



Epigenetics

Epigenetics controls the genetic information flow



Challenge

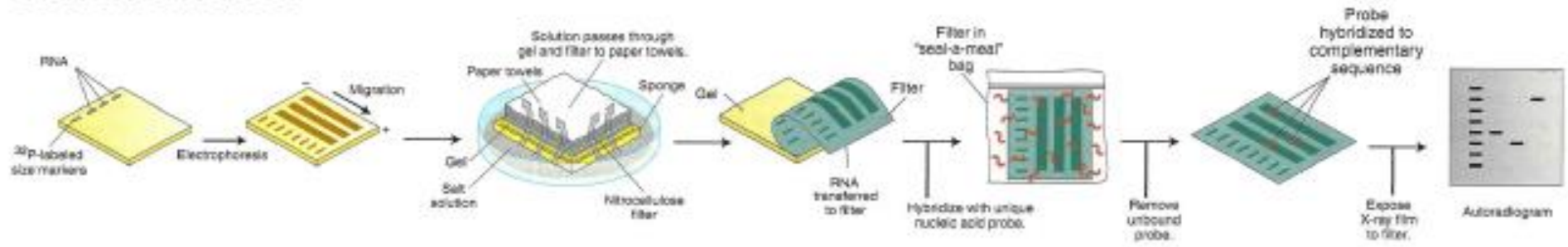
Challenge: how to measure/detect genes and their products in a massively parallel way?

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

Gene expression

How to measure the expression of genes

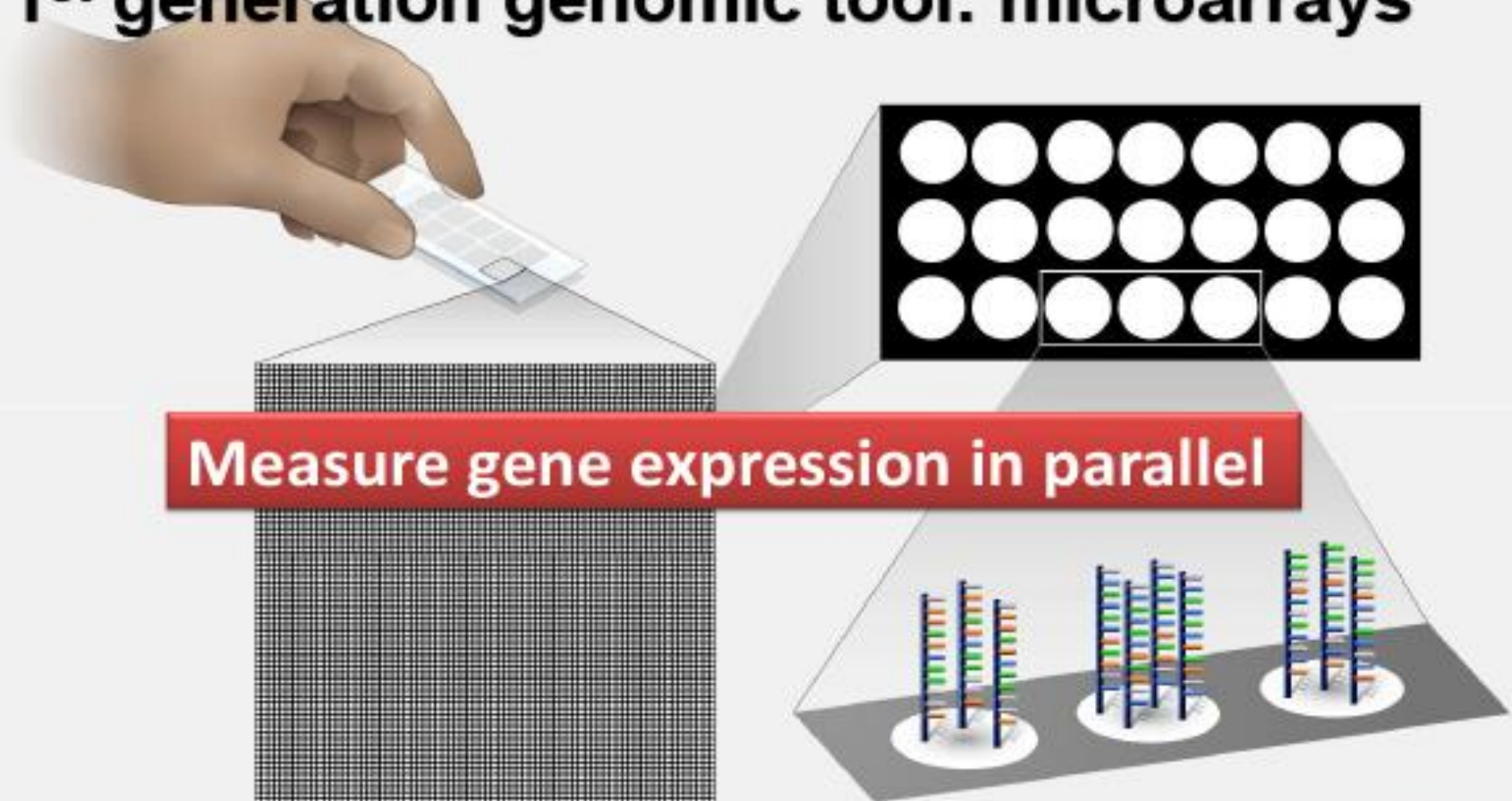
Northern blot



laborious and low throughput

Microarrays

1st generation genomic tool: microarrays

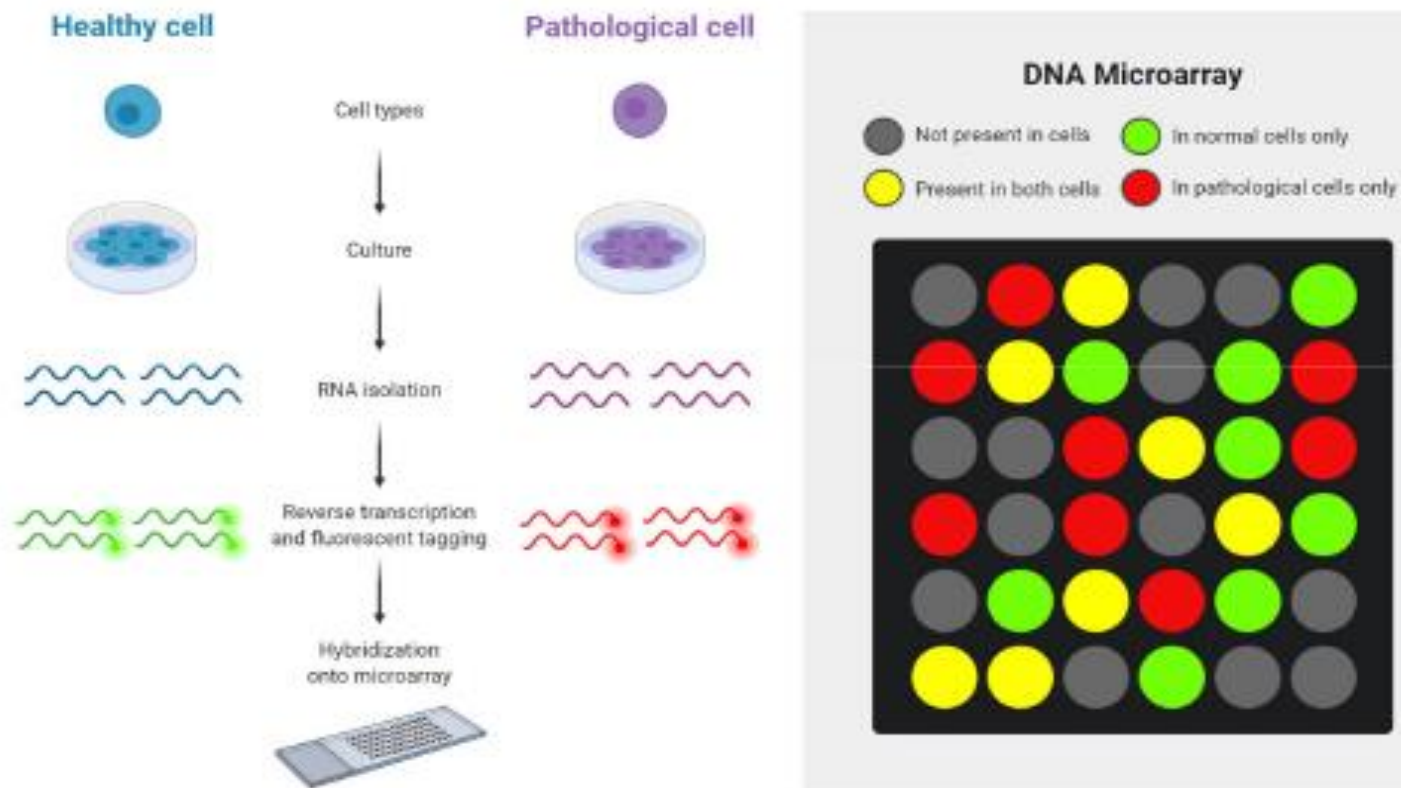


The diagram illustrates the process of a microarray experiment. A hand is shown placing a small, rectangular microarray chip onto a large, rectangular block with a fine grid pattern. To the right, a 3x7 grid of white circles is shown on a black background, with a 1x3 sub-grid highlighted by a white box. Below this, a red banner contains the text 'Measure gene expression in parallel'. At the bottom right, three DNA double helix structures are shown, each with a different color scheme (blue/green, blue/red, and blue/green), representing different gene expression profiles.

Measure gene expression in parallel

Microarrays

Microarrays – technologies of hybridization

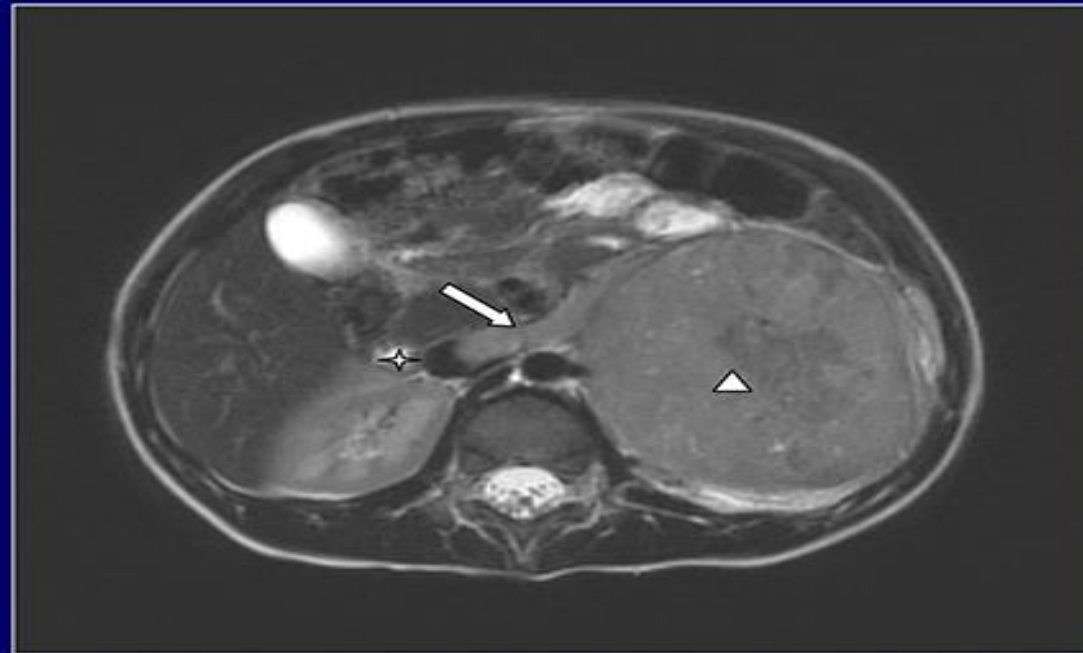


Wilms tumor

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

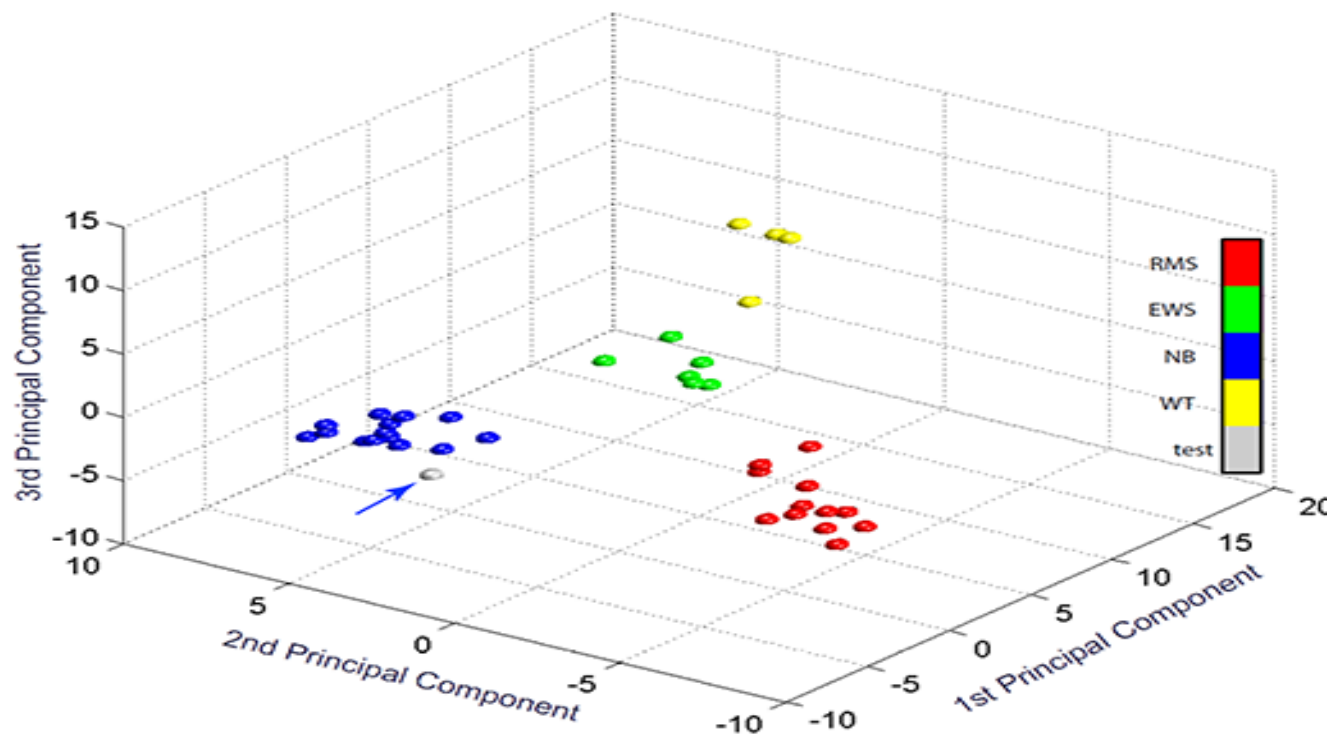


Initial diagnosis: Wilm's tumor



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

Sequencing

Modern Sequencing Technologies

First generation

Second generation
(next generation sequencing)

Third generation



Sanger Sequencing
Maxam and Gilbert
Sanger chain termination

500-1,000 bp

454, Solexa
Ion Torrent
Illumina

~50-600 bp

PacBio
Oxford Nanopore
Illumina Novaseq Series

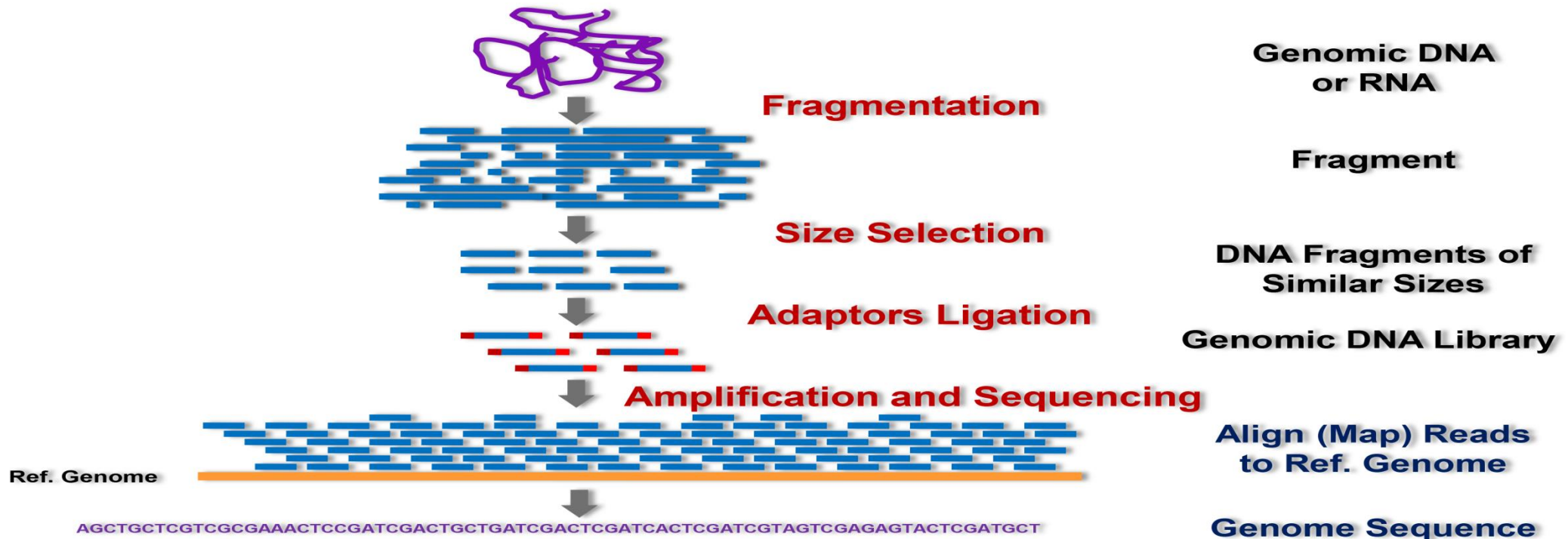
>10,000 bp

Short-read sequencing

Long-read sequencing

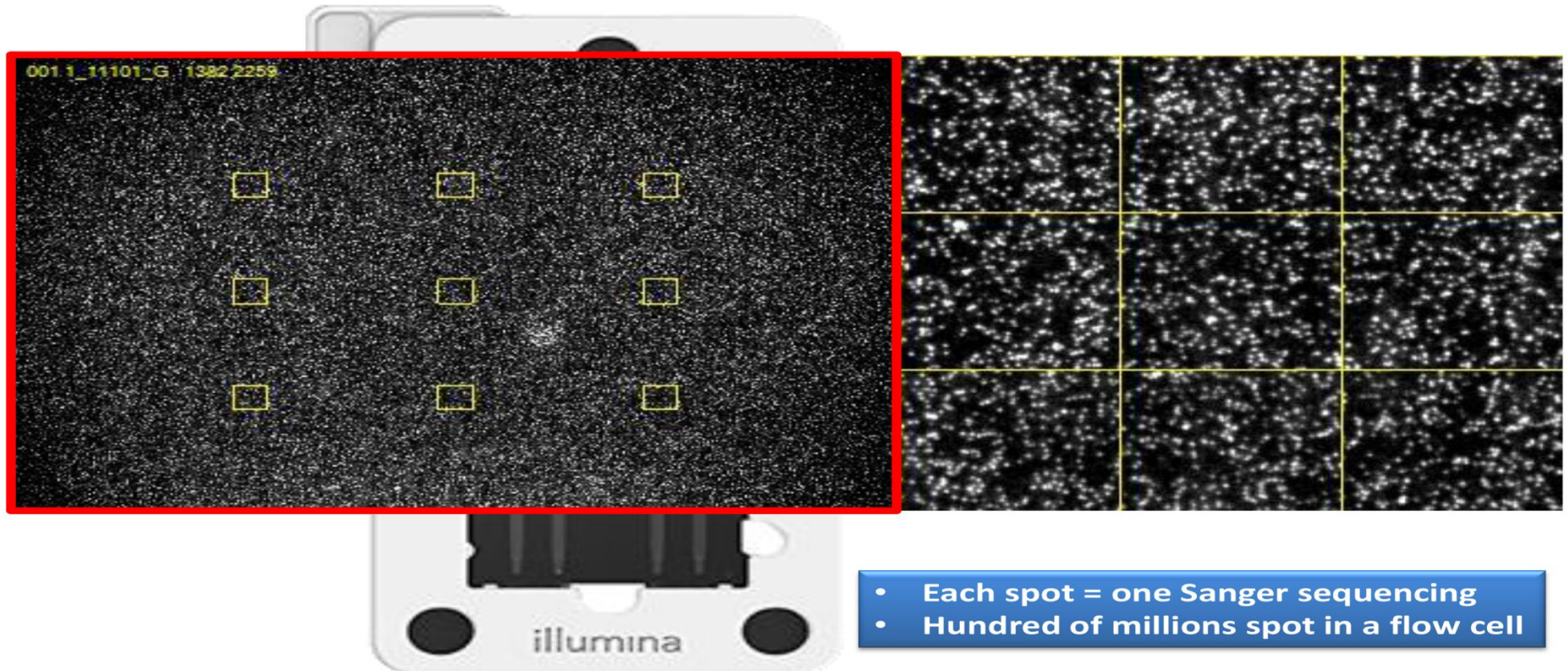
Next-generation sequencing

Next-Generation Sequencing



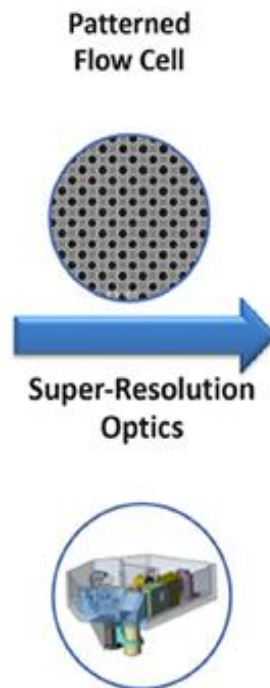
Massively Parallel Sequencing

Massively Parallel Sequencing



Desktop sequencers

Modern desktop sequencers



- 1.2 billion reads
- 400 Gb (~133 human genomes)
- <40 hours

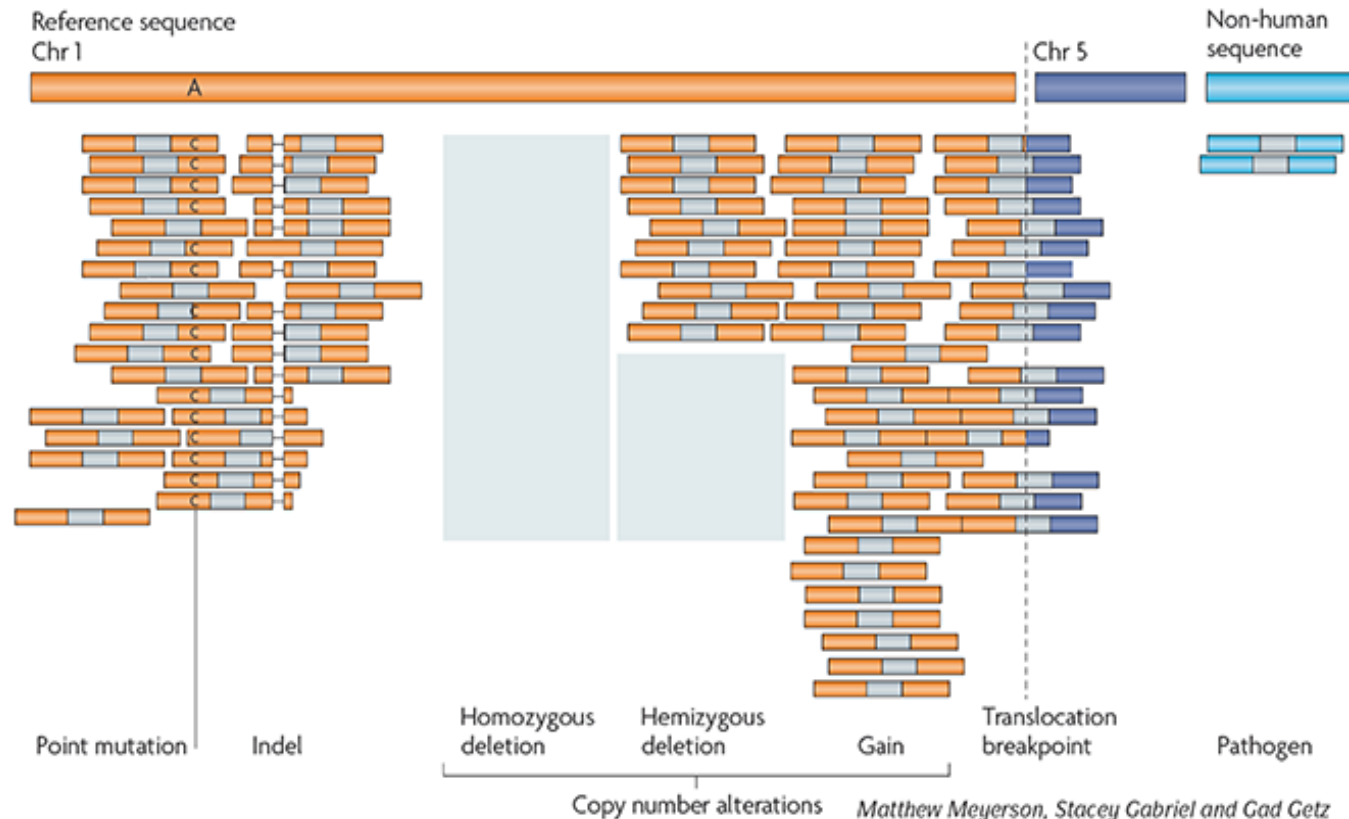
Sequencing

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

- **No prior knowledge is needed for probe design as in microarrays**
- **Massive capacity at a base-pair resolution**
 - 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 types of genomic and epigenomic information**

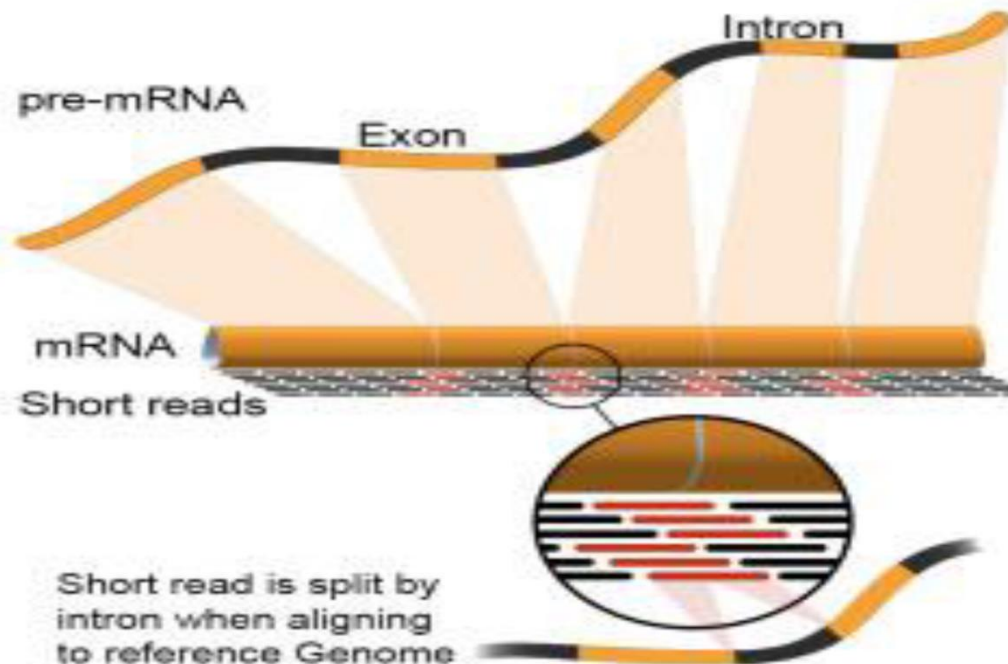
Genetic alterations

Genomic alterations detected by DNA sequencing



Genomic Alterations

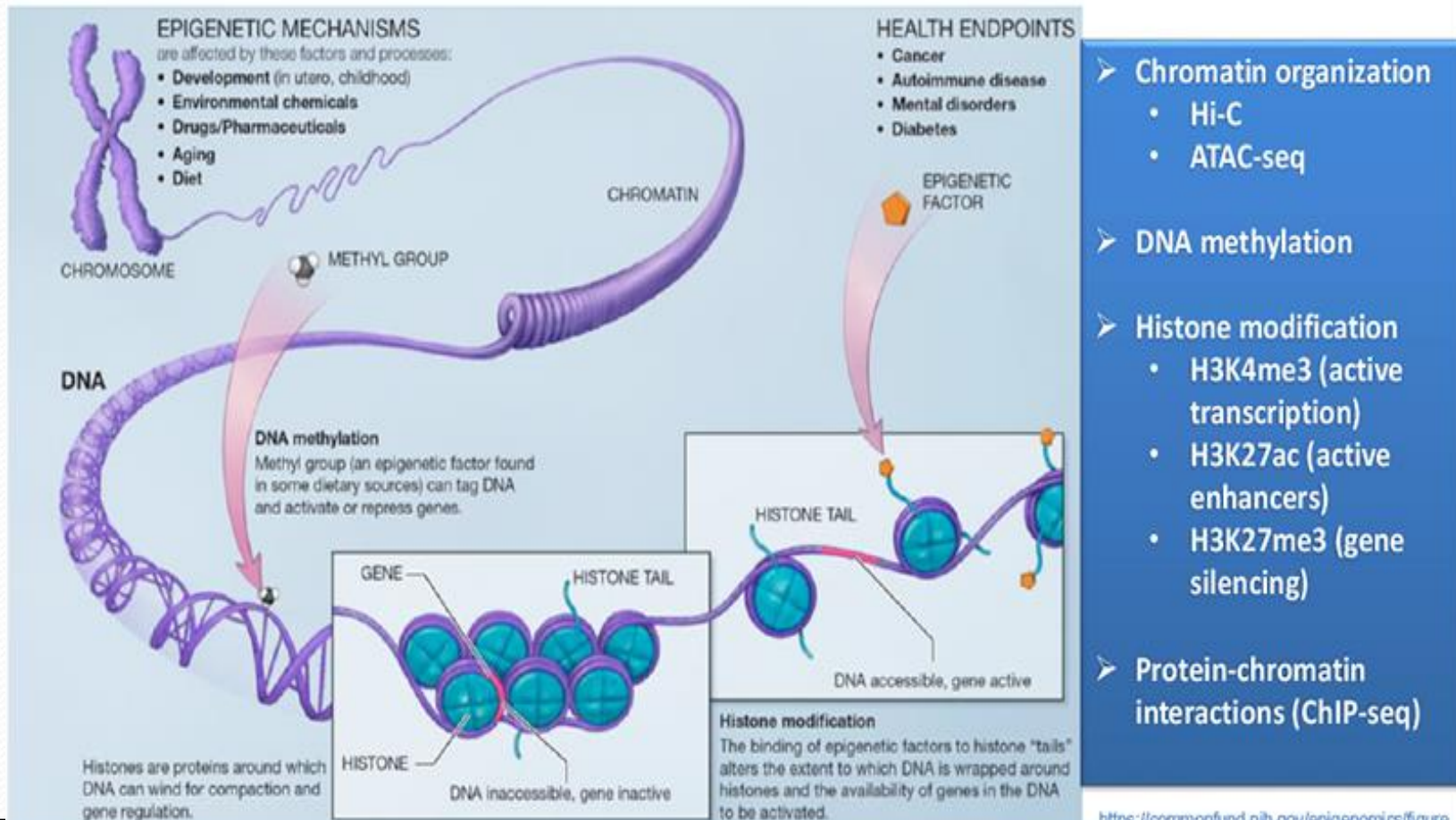
Genomic Alterations Detected by RNA Transcriptome Sequencing



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

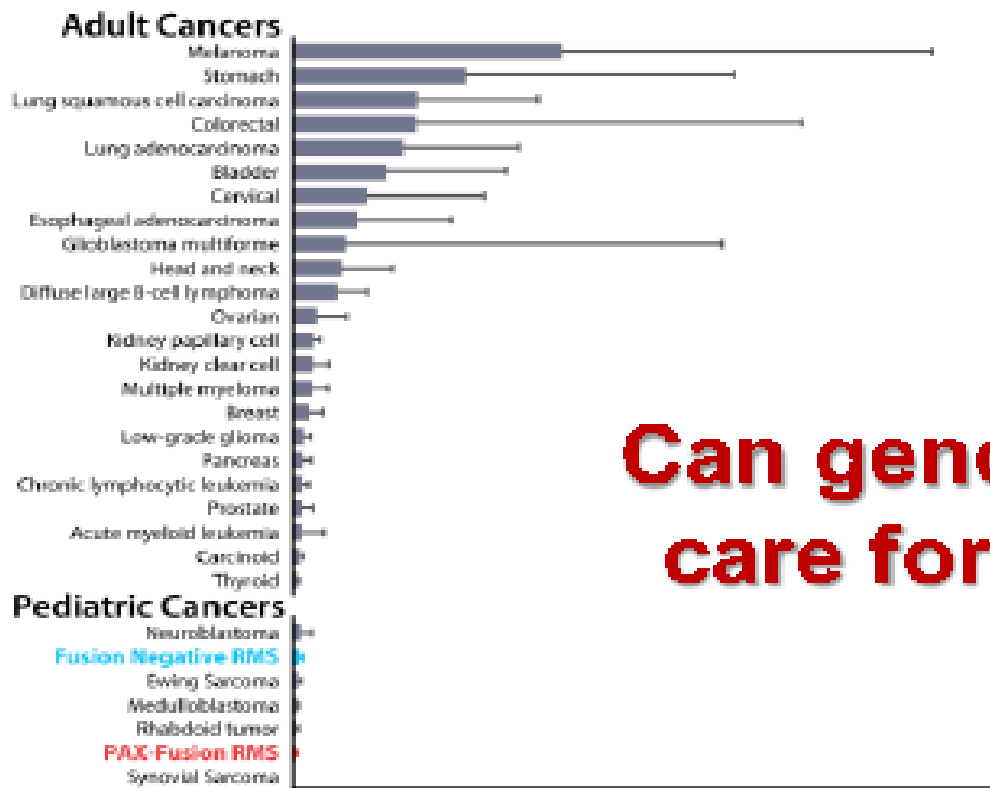
Epigenetics

Study epigenetics, the control of gene expression



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

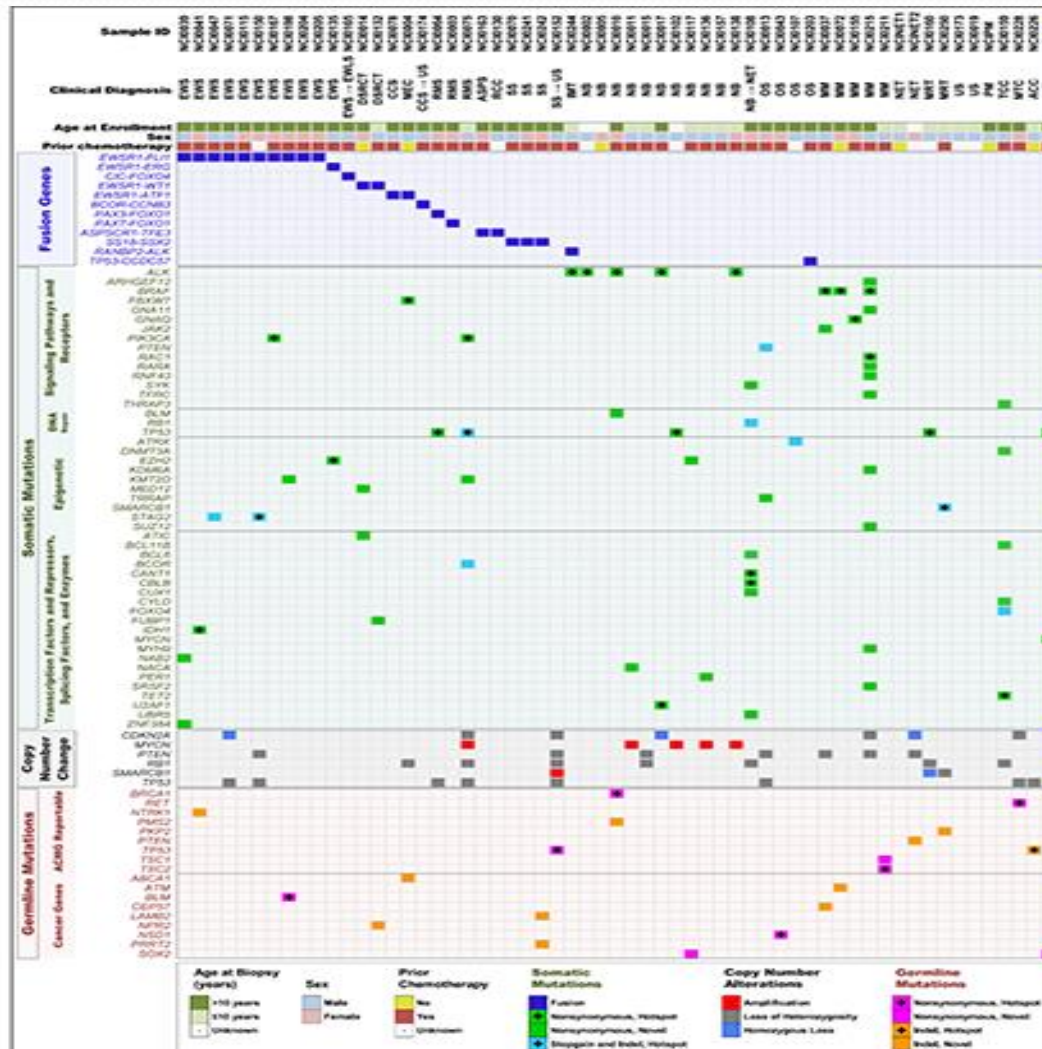
Title: “Comprehensive Omics Analysis of Pediatric Solid Tumors and Establishment of a Repository for Related Biological Studies” or Omics protocol

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 Fusion genes



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

Germline mutations

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

Table 1. Germline mutations in American College of Medical Genetics (ACMG) reportable genes and tumor suppressor genes identified in 7 patients

Sample	Diagnosis	Gene	Mutation	Disease	Hotspot	Notes	Reportable by Strict ACMG Criteria
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>	Q131X	Hereditary breast and ovarian cancer syndrome	Yes	Pathogenic, reportable	Yes
NCI0010	NB	<i>PM52</i>	p.K356fs	Lynch syndrome and mismatch repair cancer syndrome	No	Frameshift deletion of tumor suppressor gene	Yes
NCI0212	NET	<i>PTEN</i>	p.R14fs	PTEN Hamartoma tumor syndrome	No	Frameshift deletion of tumor suppressor gene	Yes
NCI0228	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_359delGinsTT	No
NCI0211	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
NCI0211	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

NOTE: Mutations were confirmed by direct visualization on an IGV viewer, and by Sanger sequencing.

Abbreviations: ACC, adrenocortical carcinoma; MM, malignant melanoma; MTC, medullary thyroid carcinoma; NET, neuroendocrine tumor; RMS, rhabdomyosarcoma; SS, synovial sarcoma; US, undifferentiated sarcoma; horizontal arrow indicates change in diagnosis.

Somatic mutations

Approximately 50% (30/59) of Pediatric and Adolescent Young Adults with Cancers Have Actionable Somatic Mutations

Table 2. Summary of actionable mutations in relapsed and refractory pediatric solid tumors

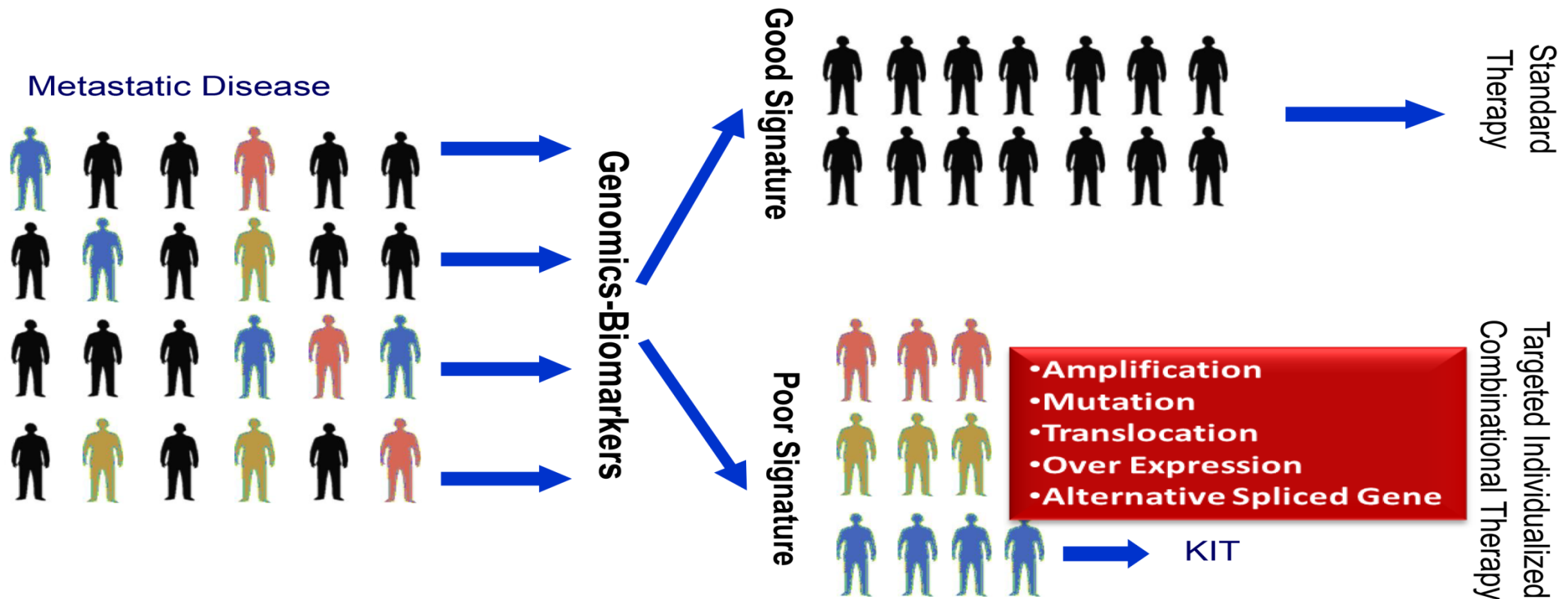
Sample	Diagnosis	Gene	Stage	Modality	Mutation	AA Change	Level	Drug	Clinical trial: Pediatric	PDA-Approval in adults	Exact mutation vs. hotspot	Reference preclinical data for level 3
NCI0037	HM	BR4F	Relapsed	WES/WTS	NS SNV	p.V600E	1	Vemurafenib, dabrafenib	Yes	Yes	Exact	—
NCI0072	HM	BR4F	Diagnostic	WES/WTS	NS SNV	p.V600E	1	Vemurafenib, dabrafenib	Yes	Yes	Exact	—
NCI0285	HM	BR4F	Relapsed	WES/WTS	NS SNV	p.V600E	1	Vemurafenib, dabrafenib	Yes	Yes	Exact	—
NCI0155	HM	GNAG	Relapsed	WES/WTS	NS SNV	p.Q209L	1	Temecicimus, trametinib, vemurafenib	No	Yes	Exact	—
NCI0002	NB	ALK	—	WES/WTS	NS SNV	p.R1275G	2a	Crizotinib	Yes	Yes	Exact	—
NCI0010	NB	ALK	Relapsed	WES/WTS	NS SNV	p.P114V	2a	Crizotinib	Yes	Yes	Exact	—
NCI0017	NB	ALK	Relapsed	WES/WTS	NS SNV	p.P114L	2a	Crizotinib	Yes	Yes	Exact	—
NCI0118	NB	ALK	Relapsed	WES/WTS	NS SNV	p.Y1278S	2a	Crizotinib	Yes	Yes	Exact	—
NCI0244	IMT	ALK	Relapsed	WTS	ANAPC2-ALK fusion	—	2a	Crizotinib	No	Yes	Exact	—
NCI0244	IMT	ALK	Relapsed	WES/WTS	NS SNV	p.Y117T	2a	Crizotinib	No	Yes	Exact	—
NCI0216	HM	GNAG	Relapsed	WES/WTS	NS SNV	p.S248P	2a	Trametinib	No	Yes	—	—
NCI0041	EWS	DNF	Relapsed	WES/WTS	NS SNV	p.R102C	2a	IDH1 inhibitors	No	No	Exact	—
NCI0075	BMS	PRSC4	Relapsed	WES/WTS	NS SNV	p.P104Q	2a	PI3K/AKT/mTOR inhibitors	Yes	Yes	Exact	—
NCI0167	EWS	PRSC4	Refractory	WES/WTS	NS SNV	p.D907G	2a	PI3K/AKT/mTOR inhibitors	Yes	Yes	Exact	—
NCI0013	OS	PTEN	Relapsed	WES/WTS	Frameshift deletion	p.R80fs	2a	PI3K/AKT/mTOR inhibitors	Yes	No	—	—
NCI0122	NET	PTEN	—	WES/WTS	Germline frameshift deletion/somatic LOH	p.R14fs	2a	PI3K/AKT/mTOR inhibitors	Yes	No	—	—
NCI0229	MTC	RET	Relapsed	WES/WTS	Germline SNV	p.M99T	2a	Vandetanib	Yes	Yes	Exact	—
NCI0017	NB	CDKN24	Relapsed	SNP Array/WTS	Homozygous loss	—	3	CDK4/6 inhibitor	No	No	—	38
NCI0071	EWS	CDKN24	Relapsed	SNP Array/WTS	Homozygous loss	—	3	CDK4/6 inhibitor	No	No	—	36
NCI0112	NET	CDKN24	—	SNP Array/WTS	Homozygous loss	—	3	CDK4/6 inhibitor	No	No	—	38
NCI0011	NB	MYCN	Relapsed	SNP Array/WTS	Amplification	—	3	Bromodomain inhibitors	No	No	—	37
NCI0176	BMS	MYCN	Relapsed	SNP Array/WTS	Amplification	—	3	Bromodomain inhibitors	No	No	—	37
NCI0102	NB	MYCN	—	SNP Array/WTS	Amplification	—	3	Bromodomain inhibitors	No	No	—	37
NCI0116	NB	MYCN	Relapsed	SNP Array/WTS	Amplification	—	3	Bromodomain inhibitors	No	No	—	37
NCI0118	NB	MYCN	Relapsed	SNP Array/WTS	Amplification	—	3	Bromodomain inhibitors	No	No	—	37
NCI0238	WT	MYCN	Relapsed	WES/WTS	NS SNV	p.P44L	3	Bromodomain inhibitors	No	No	—	37, 38
NCI0160	HRT	SMARCB1	—	SNP Array/WTS	Homozygous loss	—	3	CDK2 inhibitors	No	No	—	39, 40
NCI0260	HRT	SMARCB1	Refractory	WES/WTS	NS SNV	p.R100X	3	CDK2 inhibitors	No	No	—	39, 40
NCI0047	EWS	STAG2	Relapsed	WES/WTS	NS SNV	p.E504X	3	PARP inhibitors	Yes	No	—	41
NCI0110	EWS	STAG2	—	WES/WTS	NS SNV	p.R26X	3	PARP inhibitors	Yes	No	Hotspot	41
NCI028	HM	TSC1	Relapsed	WES/WTS	NS SNV	p.S828R	3	Everolimus	No	Yes	—	42
NCI028	HM	TSC2	Relapsed	WES/WTS	NS SNV	p.T248A	3	Everolimus	No	Yes	—	42

NOTE: SNVs were confirmed by direct visualization on an ISV viewer, and validation by Sanger sequencing or confirmation CLIA-certified laboratories.

Abbreviations: EWS, Ewing sarcoma; IMT, epithelioid inflammatory myofibroblastic sarcoma; HM, malignant melanoma; HRT, malignant rhabdoid tumor; MTC, medullary thyroid carcinoma; NB, neuroblastoma; NET, neuroendocrine tumor; OS, osteosarcoma; BMS, rhabdomyosarcoma; WT, Wilms tumor.

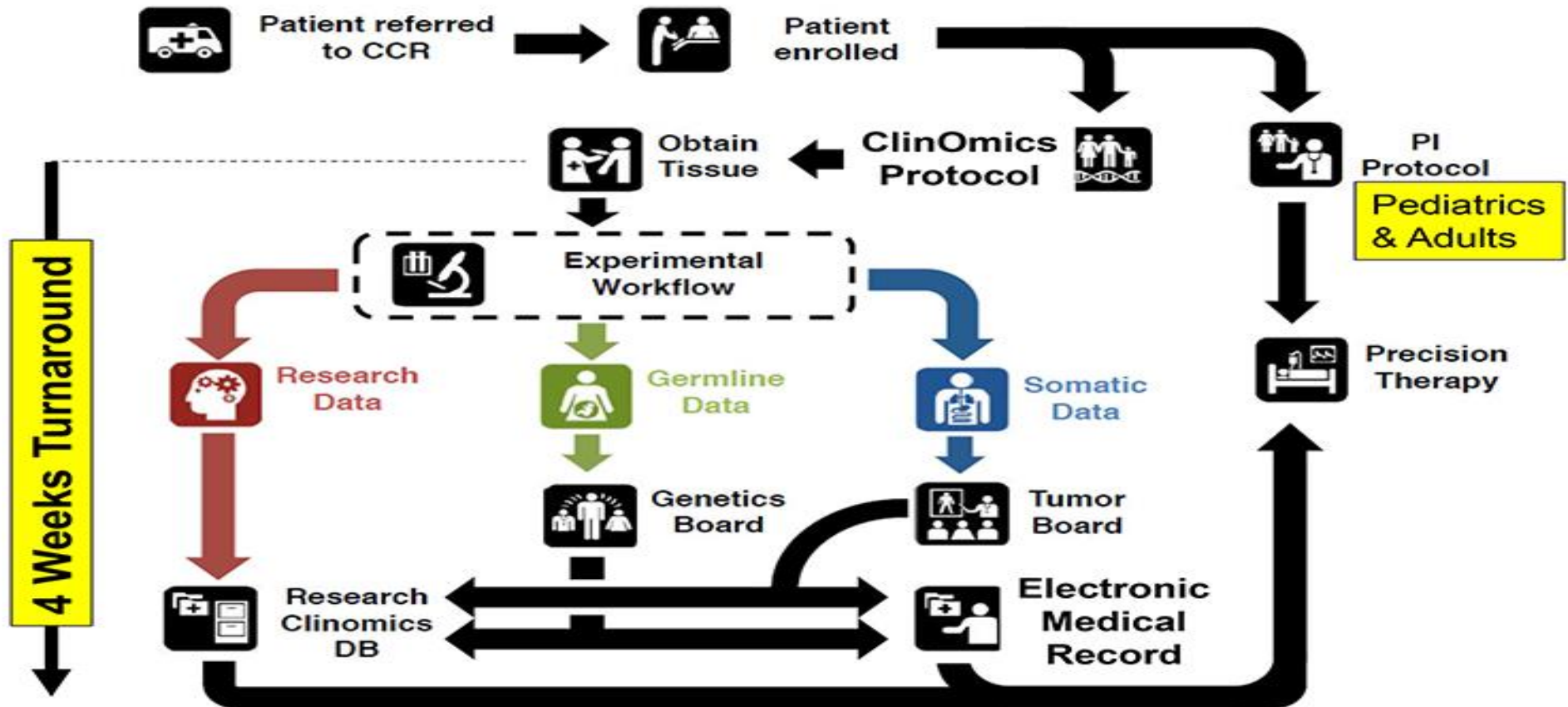
Future Trials

Genomics Enabling Precision Therapy-The Future for Pediatric Trials



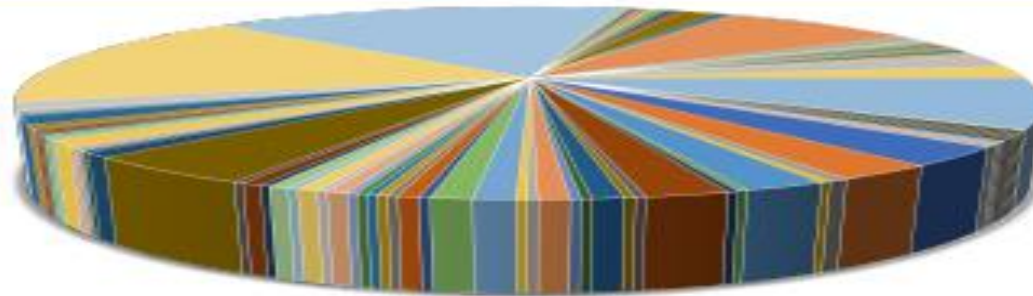
ClinOmics program

CCR ClinOmics Program-CLIA



Patient diagnoses

396 Patients of 93 diagnoses



BRAC

- (1) Anaplastic Astrocytoma
- (2) Anaplastic Piloc
- (3) Bladder cancer
- (4) Cholangiocarcinoma
- (5) Dermatofibrosarcoma protuberans
- (6) Diffuse intrinsic pontine glioma
- (7) Ependymoma
- (8) Glioblastoma
- (9) Glioblastoma
- (10) Grade 3 Oligodendroglioma
- (11) Invasive well differentiated squamous cell carcinoma
- (12) Lymphocytoma
- (13) Melanoma
- (14) Mesothelioma Pleural
- (15) Metastatic Pancreatic Neuroendocrine Carcinoma
- (16) Multiple Brain 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 Spinal meningioma
- (27) Anaplastic Fibrous Histiocytoma
- (28) Breast cancer
- (29) Chondroma
- (30) Desmoplastic Fibrosarcoma
- (31) Endometrial cancer
- (32) Ewing's sarcoma
- (33) Glioblastoma
- (34) Hepatic Angiosarcoma
- (35) Keratoacanthoma
- (36) Melanocytoma
- (37) Merkel Cell Carcinoma
- (38) Mesothelioma Visceral Pleural
- (39) MPM2
- (40) Myxopapillary Ependymoma
- (41) Neuronal tumor
- (42) Ovarian Serous Carcinoma
- (43) Pilocytic Astrocytoma
- (44) Prostate cancer
- (45) Pseudopapillary carcinoma
- (46) Small Cell Carcinoma of the ovary hypercalcemic type (OEDHE)
- (47) Teratoma

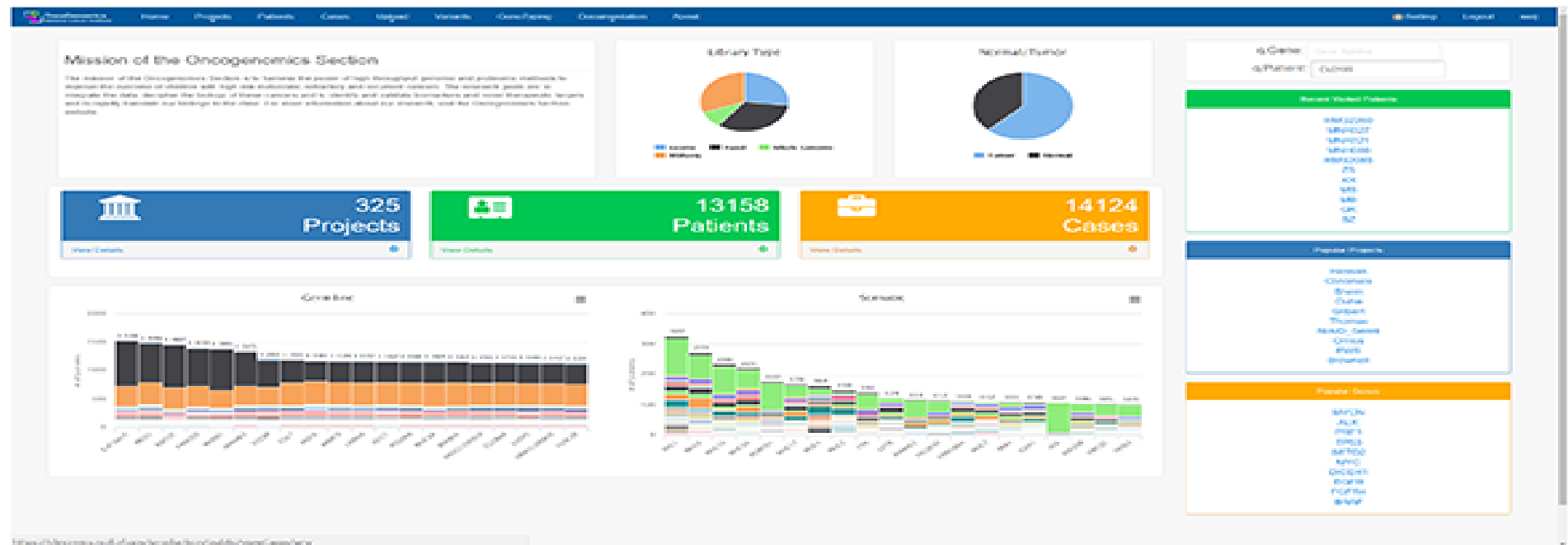
- (48) Acute myeloid leukemia
- (49) Anaplastic meningioma
- (50) Astrocytoma
- (51) Carcinoid, BRCA1 positive
- (52) Clear cell sarcoma
- (53) Desmoplastic small round cell tumor
- (54) Endometrial Stromal Sarcoma
- (55) Extracranial Small Cell Cancer
- (56) Glioma
- (57) Hepatocellular cancer
- (58) Left Cerebellar Sarcoma
- (59) Medullary Thyroid Cancer metastatic
- (60) Mesothelioma
- (61) Metastatic Acid Carcinoma
- (62) Multinodular and Circulating Neuronal Tumor
- (63) Neuroendocrine carcinoma
- (64) Neuroendocrine giant cell tumor
- (65) Ovarian Teratoma
- (66) Pilocytic Astrocytoma
- (67) Pleomorphic Xanthoastrocytoma
- (68) Pleomorphic gliomatous tumor
- (69) RCC
- (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) Ependymoma
- (79) Infiltrative cancer
- (80) Gliosarcoma
- (81) Hepatocellular carcinoma
- (82) Lung Adenocarcinoma
- (83) Medulloblastoma
- (84) Mesothelioma Peritoneal
- (85) Metastatic MEL
- (86) Multiple carcinomas
- (87) Neuroendocrine Tumor
- (88) NSCLC
- (89) Pancreatic cancer
- (90) Pleomorphic xanthoastrocytoma
- (91) Recurrent Medulloblastoma
- (92) Small cell bladder
- (93) Synovial sarcoma
- (94) Undifferentiated sarcoma

ClinOmics Data Portal

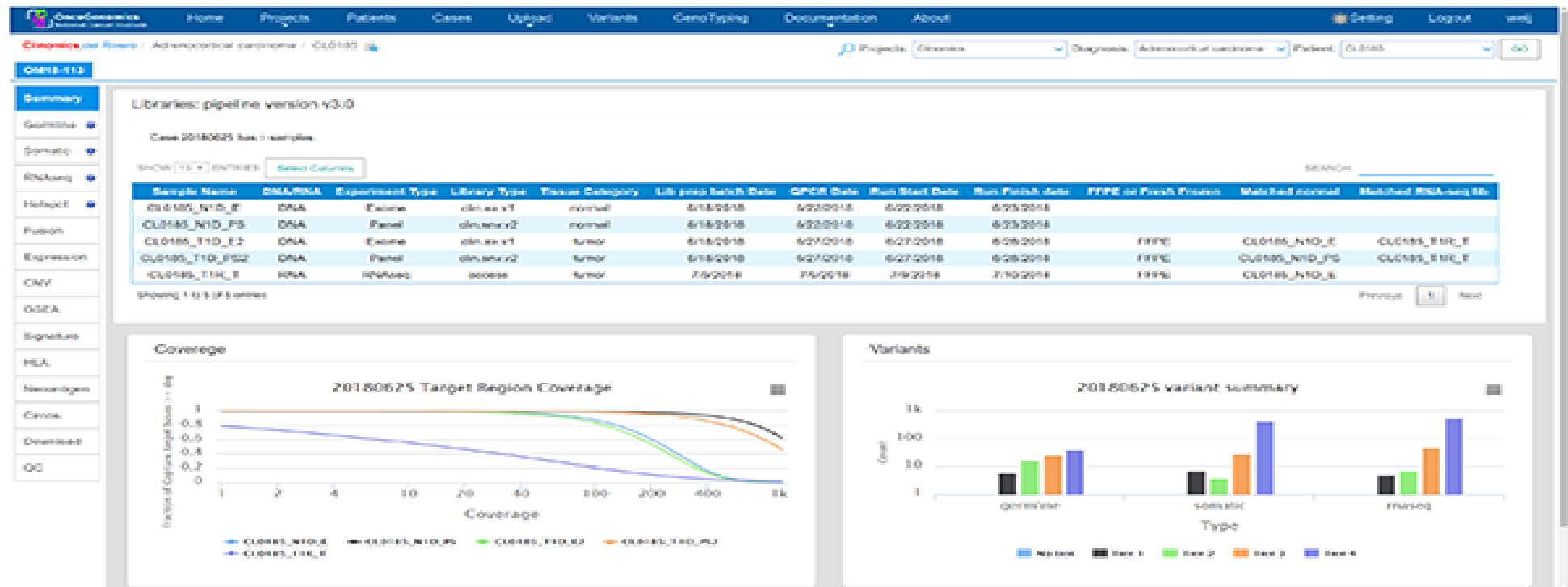
ClinOmics Data Portal

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



Patient Summary

Patient Summary Page



QC report

QC Report: Sequencing Statistics & Genotyping

Run Statistics

Summary	Close	Coverage	Transcript Coverage	Hotspot	Contours	Compare	DNA GC	RNA GC	RNA GC v2	FASTQC	Genotyping	Versions
Summary												
Genome												
Genetic												
RNAseq												
Hotspot												
Fusion												
Expression												
CRV												
GSEA												
Signature												

Showing 1 to 4 of 4 entries

100% threshold

Genotyping

Summary	Close	Coverage	Transcript Coverage	Hotspot	Contours	Compare	DNA GC	RNA GC	RNA GC v2	FASTQC	Genotyping	Versions
Summary												
Genome												
Genetic												
RNAseq												
Hotspot												
Fusion												
Expression												
CRV												
GSEA												

Showing 1 to 4 of 4 entries

100% threshold

Germline and Somatic Mutations

The screenshot displays the Ensembl genome browser interface. At the top, there is a navigation bar with links to Home, Genes, Proteins, Genes, and other genomic data. Below this, a search bar is visible. The left sidebar shows the 'Genes' track with a list of genes. The main content area shows the 'BRCA1' gene track with a detailed view of the gene structure and associated genomic data. The bottom section shows a list of genomic features with columns for Feature, Location, and other details.

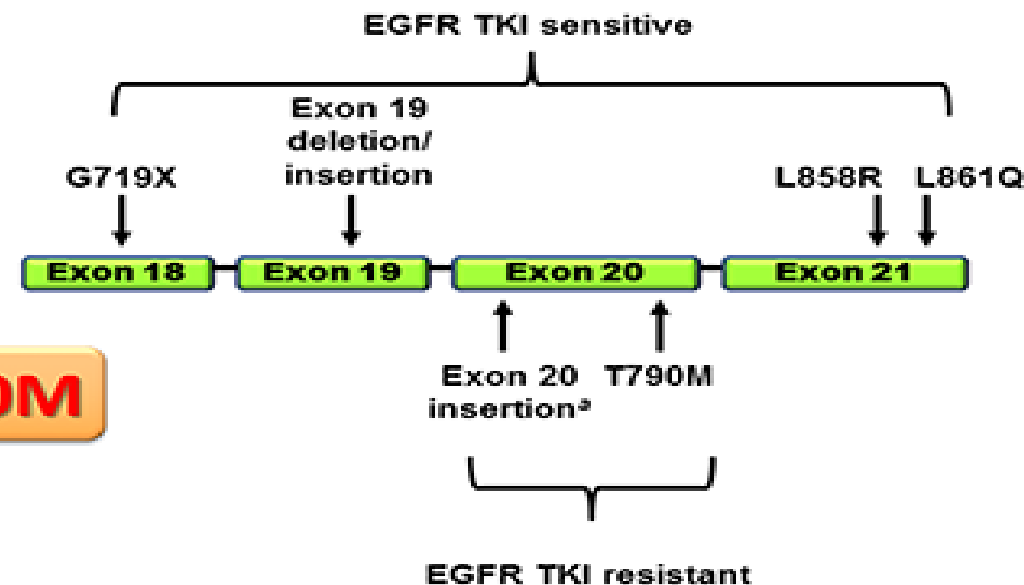
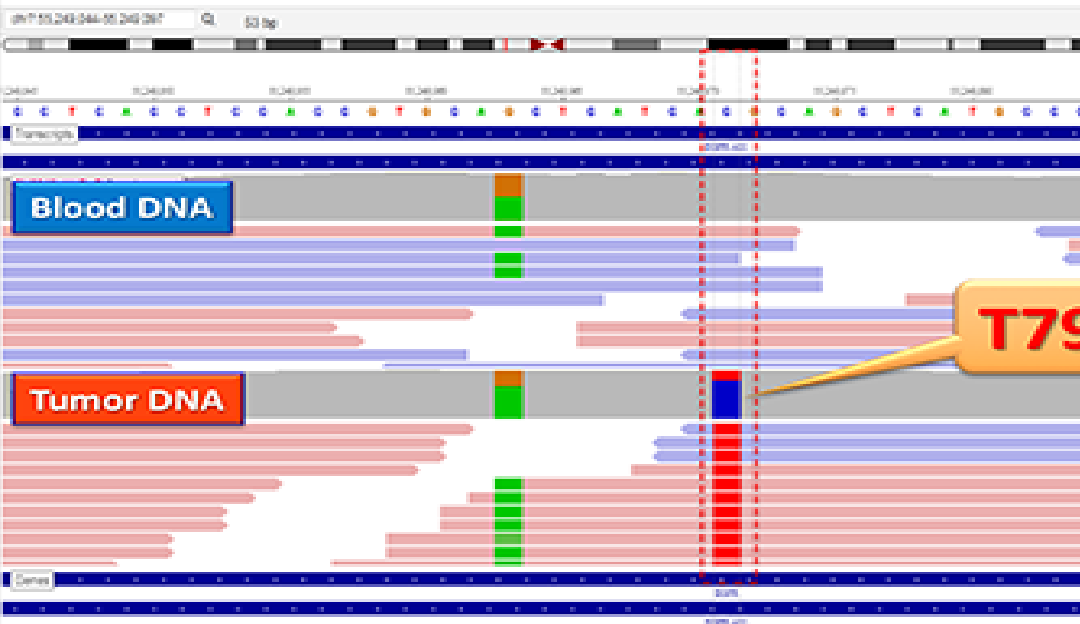
EGFR mutations

EGFR mutations in NSCLC

SV view of patient: CL0040 case: OM18-007 Total 4 sample(s)

ic (check sample to load)

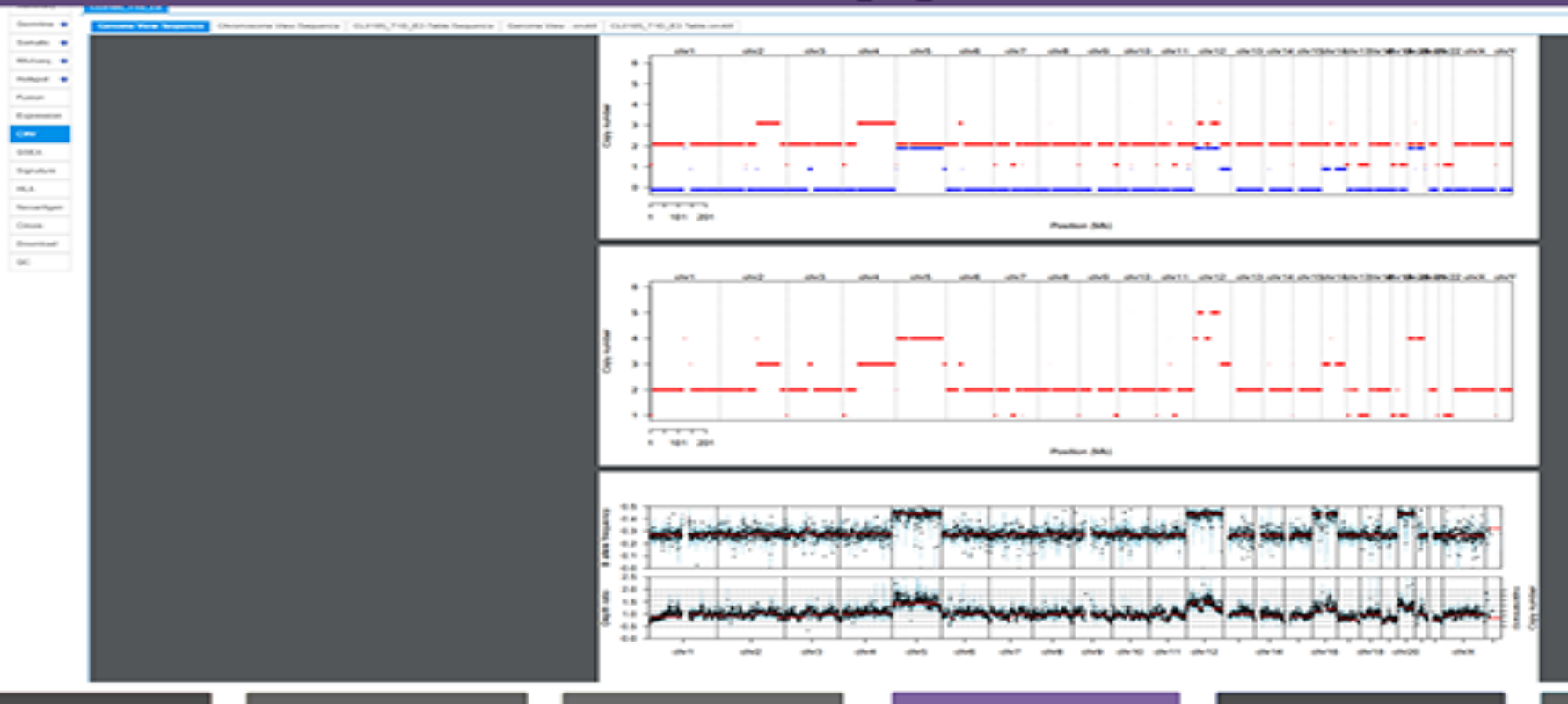
CL0040_T10_E Exome, normal (Exome, normal) CL0040_T10_E Exome, tumor (Exome, tumor) CL0040_T10_P Panel, normal (Panel, normal) CL0040_T10_P Panel, tumor (Panel, tumor)



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

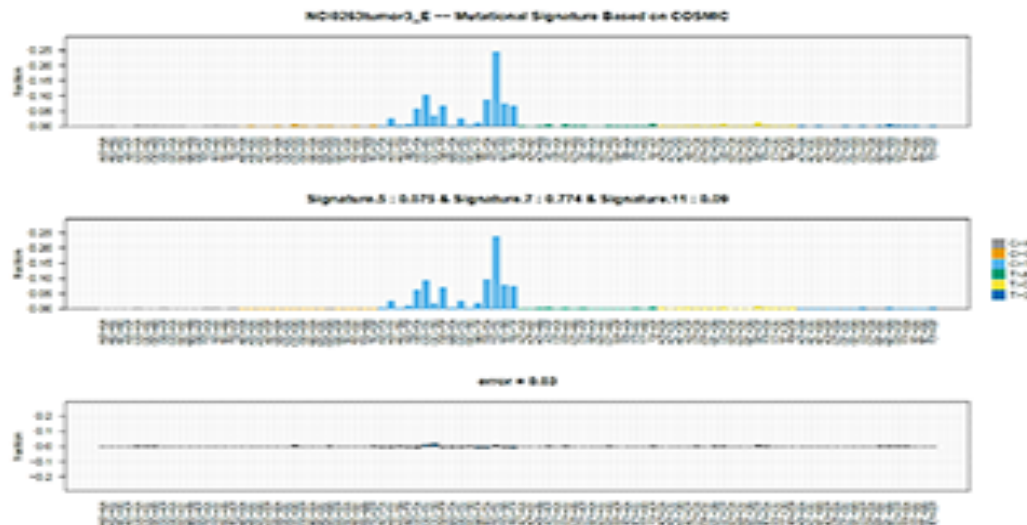
Tumor Copy Number

Tumor Copy Number

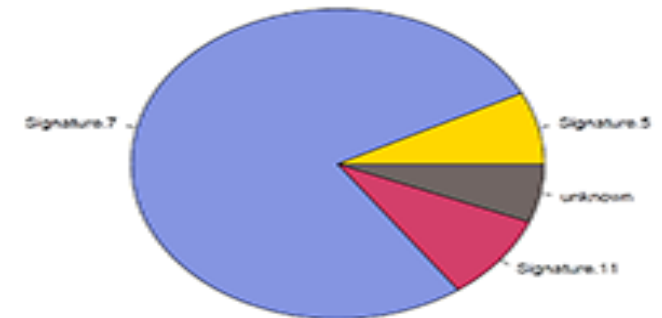


Mutation Signatures

Mutation Signatures for Tumor

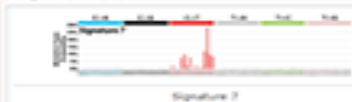


NCI0263: Melanoma



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 (viz., by formation of pyrimidine-pyrimidine photodimers) 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 Río' / 'Adrenocortical carcinoma' / 'CL0185'. Search filters are set to 'Projects: Clinomics', 'Diagnosis: Adrenocortical carcinoma', and 'Patient: CL0185'. The left sidebar lists 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). The 'Callers' dropdown is set to 'MuTect'. A 'Select Columns' button is present. The table shows 2 records out of 6 total entries, filtered from 6 total entries. The table columns are 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**

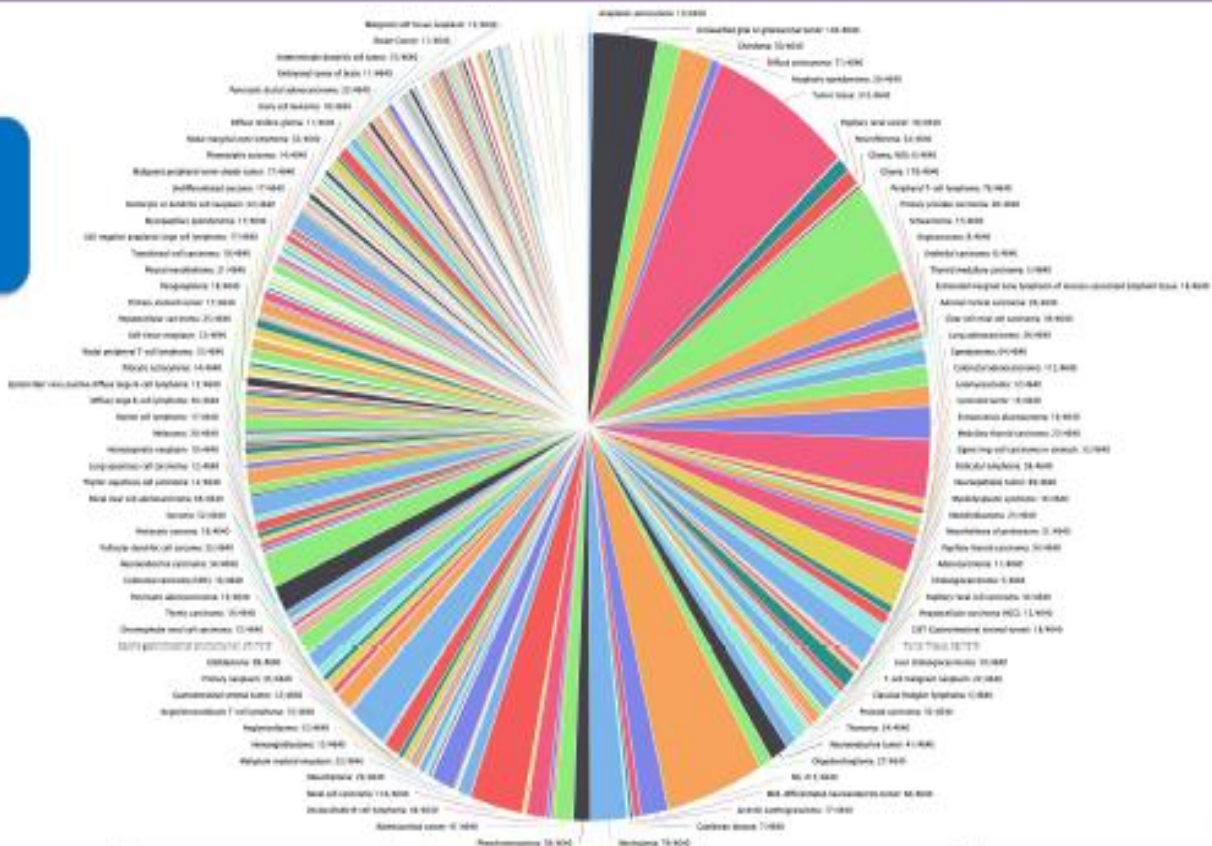
COMPASS

COMPASS Program (LP, 2019-)

11/1/2023:

Patients = 4640

Diagnosis = 496



Conclusions

Conclusions

- Next-generation sequencing is an important genomic tool to study the genomics and epigenetics of tumors
- Genomic research has significantly advanced our understanding of human cancers
- Routine integrated omics analyses of patient tumors can pinpoint rational molecular targets to improve the outcomes of childhood cancers

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