# จีโนมิกส์ประเทศไทย: การแพทย์จีโนมิกส์เพื่อการยกระดับคุณภาพชีวิตคนไทย

## Genomics Thailand: Enhancing our Quality of Life via Genomic Medicine

วันที่ 30 มีนาคม 2566 เวลา 09.30 – 16.00 น. อาคารสราญวิทย์ อุทยานวิทยาศาสตร์ประเทศไทย

NAC2023 18th NSTDA Annual Conference การประชุมวิชาการประจำปี สวทช. ครั้งที่ ๑๘

## Update State of the Art Genomic Platforms

### 15.15 – 16.00 น.

**นพ.วีรยุทธ ประพันธ์พจน์**

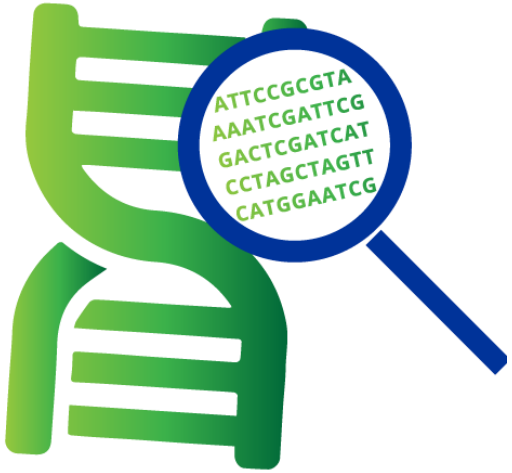
บริษัท ศูนย์พันธุศาสตร์การแพทย์ จำกัด

**อ.ดร.ธิดาทิพย์ วงศ์สุรวัฒน์**

คณะแพทยศาสตร์ศิริราชพยาบาล มหาวิทยาลัยมหิดล

**นายเฉลิมพล ศรีจอมทอง**

ศูนย์ความเป็นเลิศทางการแพทย์ด้านเวชพันธุศาสตร์
โรงพยาบาลจุฬาลงกรณ์ สภากาชาดไทย




**Q&A 10 นาที**

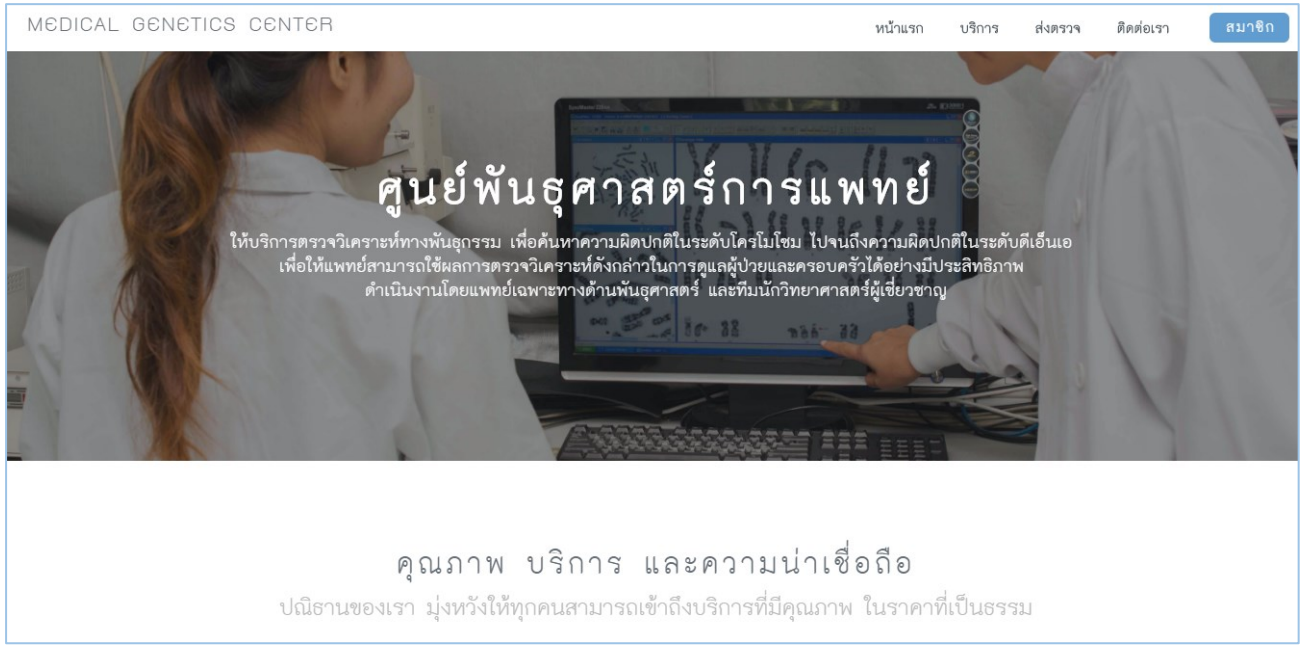
# Next generation sequencing

## current applications and future perspective

นพ.วีรยุทธ ประพันธ์พจน์, พบ.วว . (กุมารเวชศาสตร์)

American Board of Medical Genetics (Cytogenetics)

6/6 ม.6
MGC
excellence
ศูนย์พันธุศาสตร์การแพทย์
MEDICAL GENETICS CENTER


MEDICAL GENETICS CENTER
หน้าแรก
บริการ
ส่งตรวจ
ติดต่อเรา
สมาชิก
ศูนย์พันธุศาสตร์การแพทย์
ให้บริการตรวจวิเคราะห์ทางพันธุกรรม เพื่อค้นหาความผิดปกติในระดับโครโมโซม ไปจนถึงความผิดปกติในระดับดีเอ็นเอ
เพื่อให้แพทย์สามารถใช้ผลการตรวจวิเคราะห์ดังกล่าวในการดูแลผู้ป่วยและครอบครัวได้อย่างมีประสิทธิภาพ
ดำเนินงานโดยแพทย์เฉพาะทางด้านพันธุศาสตร์ และทีมนักวิทยาศาสตร์ผู้เชี่ยวชาญ
คุณภาพ บริการ และความน่าเชื่อถือ
ปณิธานของเรา มุ่งหวังให้ทุกคนสามารถเข้าถึงบริการที่มีคุณภาพ ในราคาที่เป็นธรรม

## 1 Sanger Sequencing

**Slow but good base and accurate structure**

Reads each single base one at a time

## 2 Amplified Molecule Sequencing

**Fast reads but only good base accuracy**

Reads 300 base long fragments

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10x GENOMICS

PACBIO

Oxford NANOPORE technologies

## 4 New Technologies

**Fast reads with good base and structural accuracy**

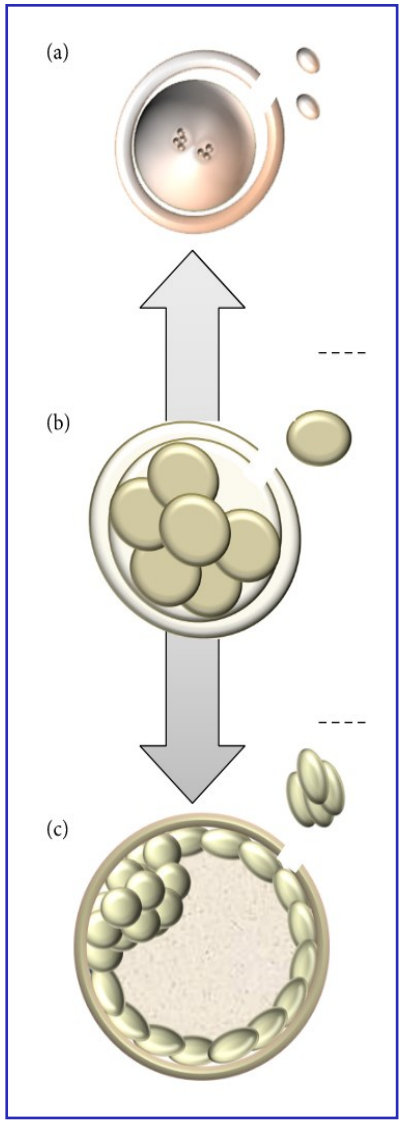
Reads 100,000 base long fragments

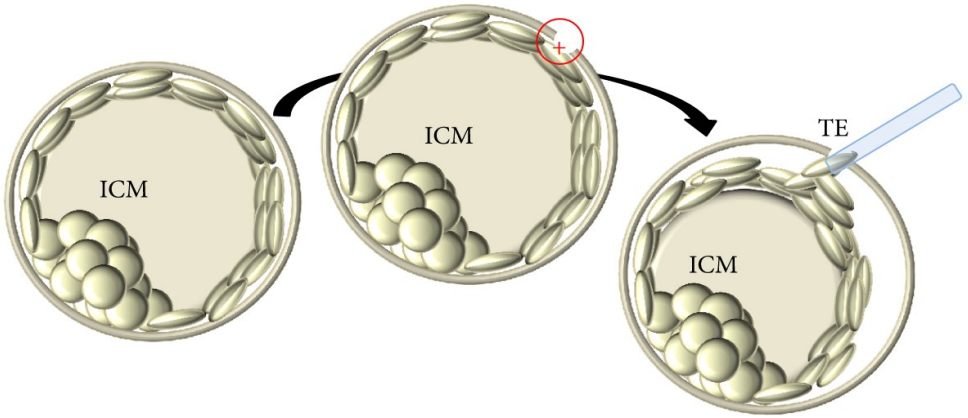
**New Technologies in Development**

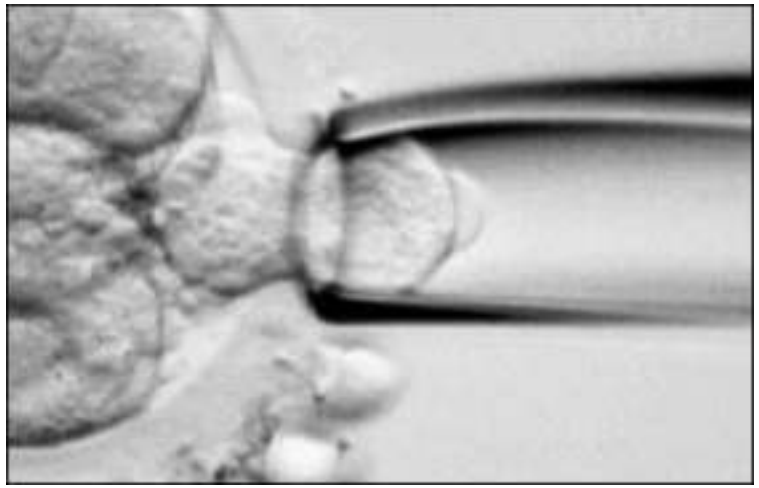
https://revolugen.co.uk/market-applications/dna-sequencing.html

# Current applications

https://static.vecteezy.com/

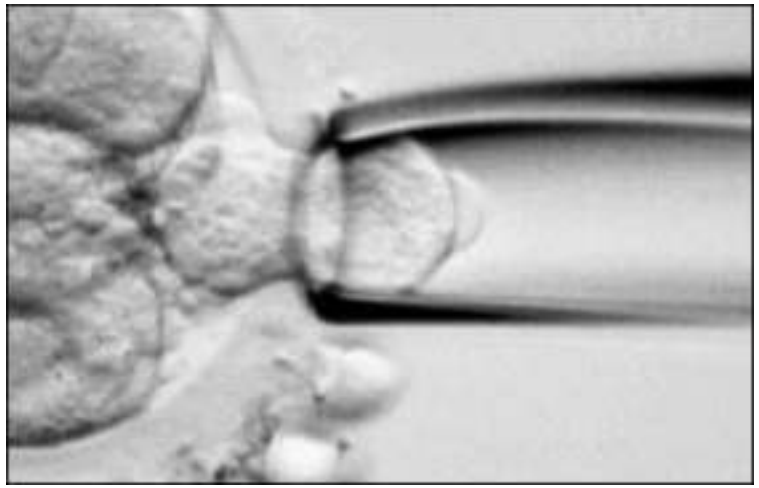


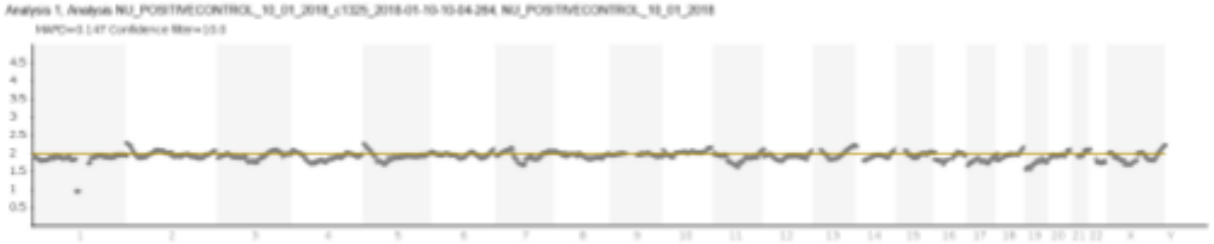



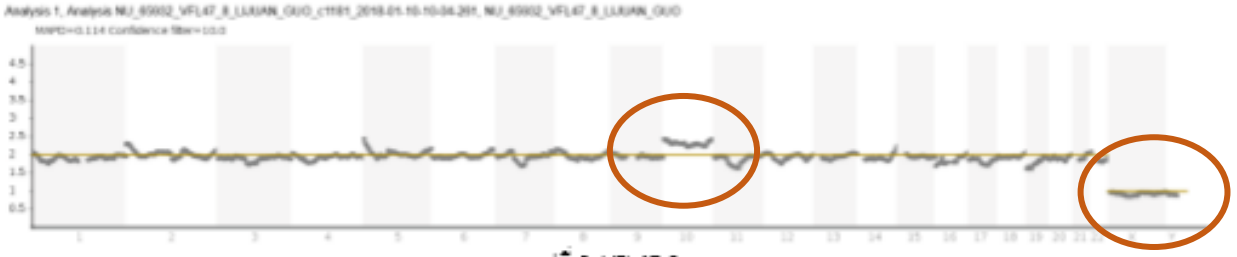
BioMed Research International
Volume 2016 (2016), Article ID 7193075
http://dx.doi.org/10.1155/2016/7193075

# Whole genome sequencing & copy number analysis

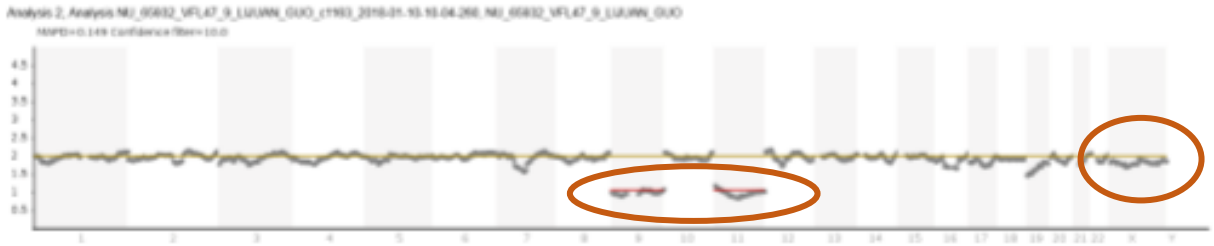




รูปที่ 1: Control



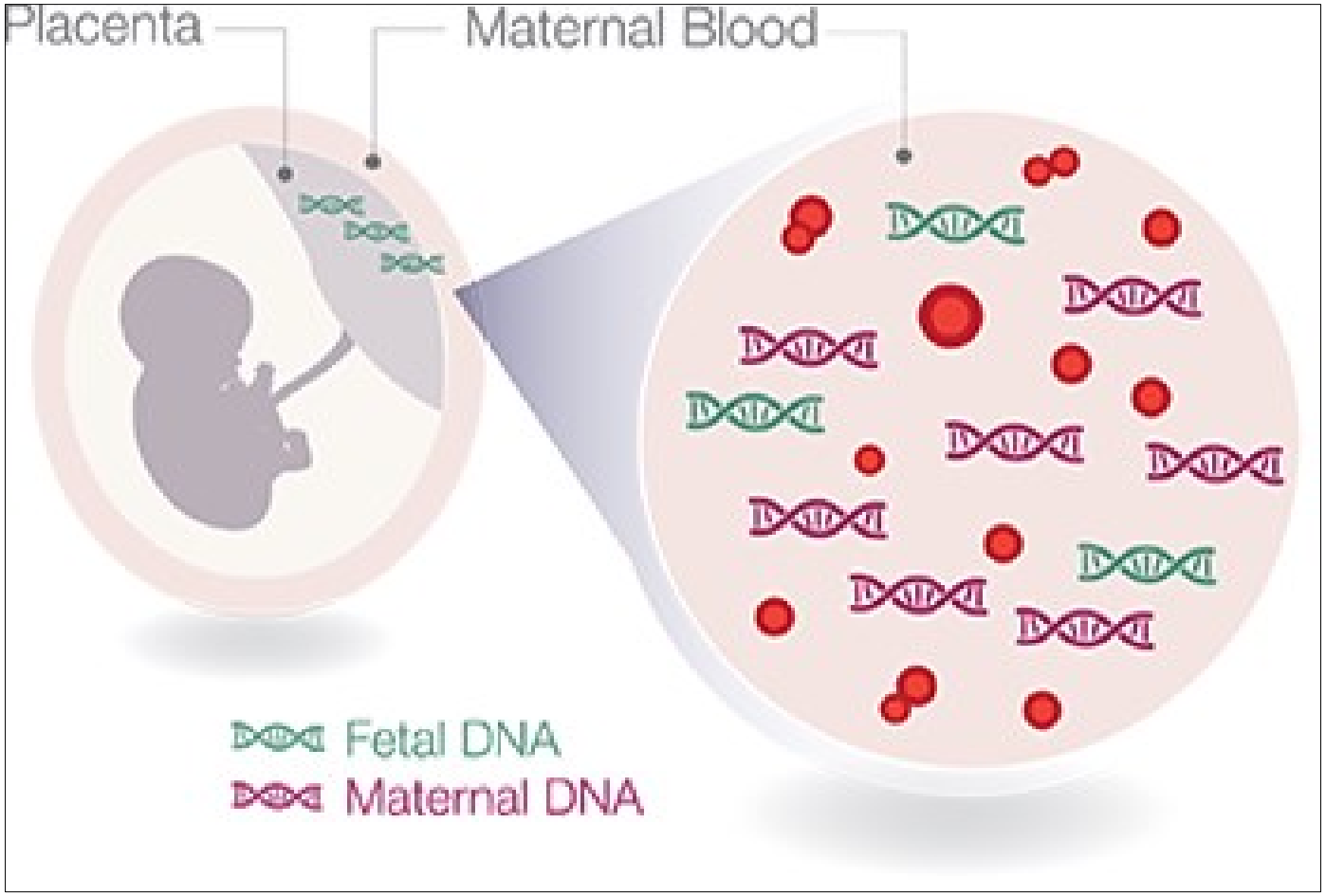
รูปที่ 2: VFL47-8



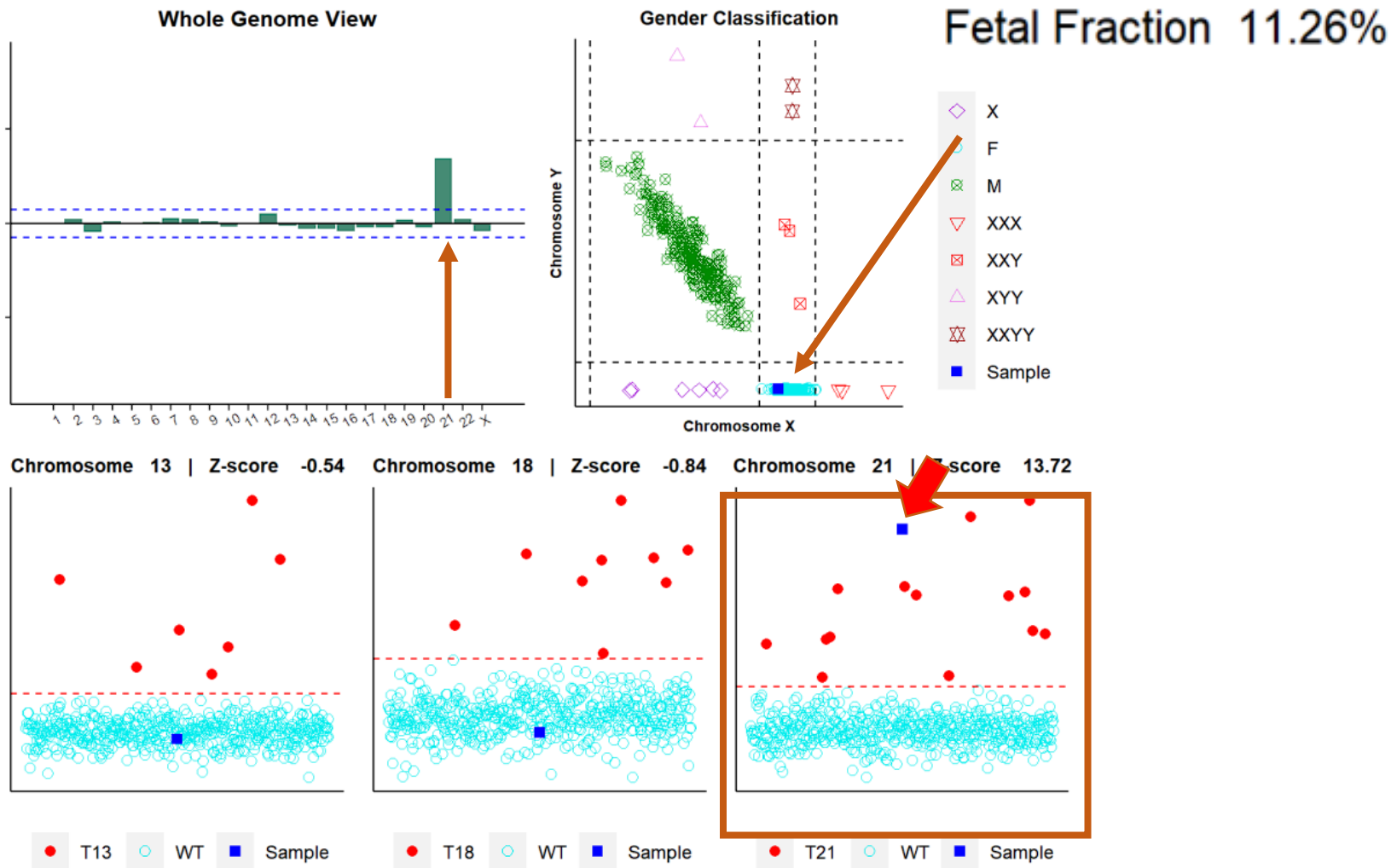
รูปที่ 3: VFL47-9

https://static.vecteezy.com/

# Non-invasive prenatal screening : NIPS



# Whole genome sequencing & chromosome counting



https://static.vecteezy.com/

# Whole exome sequencing : WES

Noonan Syndrome Association

https://www.noonansyndrome.org.uk/

# Whole exome sequencing : WES

**Noonan syndrome - PS163950 - 16 Entries**

| Location | Phenotype | Inheritance | Phenotype mapping key | Phenotype MIM number | Gene/Locus | Gene/Locus MIM number |
|---|---|---|---|---|---|---|
| 1p13.2 | Noonan syndrome 6 | AD | 3 | 613224 | NRAS | 164790 |
| 1q22 | Noonan syndrome 8 | AD | 3 | 615355 | RIT1 | 609591 |
| 2p23.2 | Noonan syndrome-like disorder with loose anagen hair 2 | AD | 3 | 617506 | PPP1CB | 600590 |
| 2p22.1 | Noonan syndrome 4 | AD | 3 | 610733 | SOS1 | 182530 |
| 2p14 | Noonan syndrome 14 | AR | 3 | 619745 | SPRED2 | 609292 |
| 3p25.2 | Noonan syndrome 5 | AD | 3 | 611553 | RAF1 | 164760 |
| 3q22.3 | Noonan syndrome 11 | AD | 3 | 618499 | MRAS | 608435 |
| 7q34 | Noonan syndrome 7 | AD | 3 | 613706 | BRAF | 164757 |
| 10q25.2 | Noonan syndrome-like with loose anagen hair 1 | AD | 3 | 607721 | SHOC2 | 602775 |
| 11p15.2 | Noonan syndrome 12 | AD | 3 | 618624 | RRAS2 | 600098 |
| 12p12.1 | Noonan syndrome 3 | AD | 3 | 609942 | KRAS | 190070 |
| 12q24.13 | Noonan syndrome 1 | AD | 3 | 163950 | PTPN11 | 176876 |
| 14q21.3 | Noonan syndrome 9 | AD | 3 | 616559 | SOS2 | 601247 |
| 22q11.21 | Noonan syndrome 2 | AR | 3 | 605275 | LZTR1 | 600574 |
| 22q11.21 | Noonan syndrome 10 | AD | 3 | 616564 | LZTR1 | 600574 |
| 22q11.22 | Noonan syndrome 13 | AD | 3 | 619087 | MAPK1 | 176948 |

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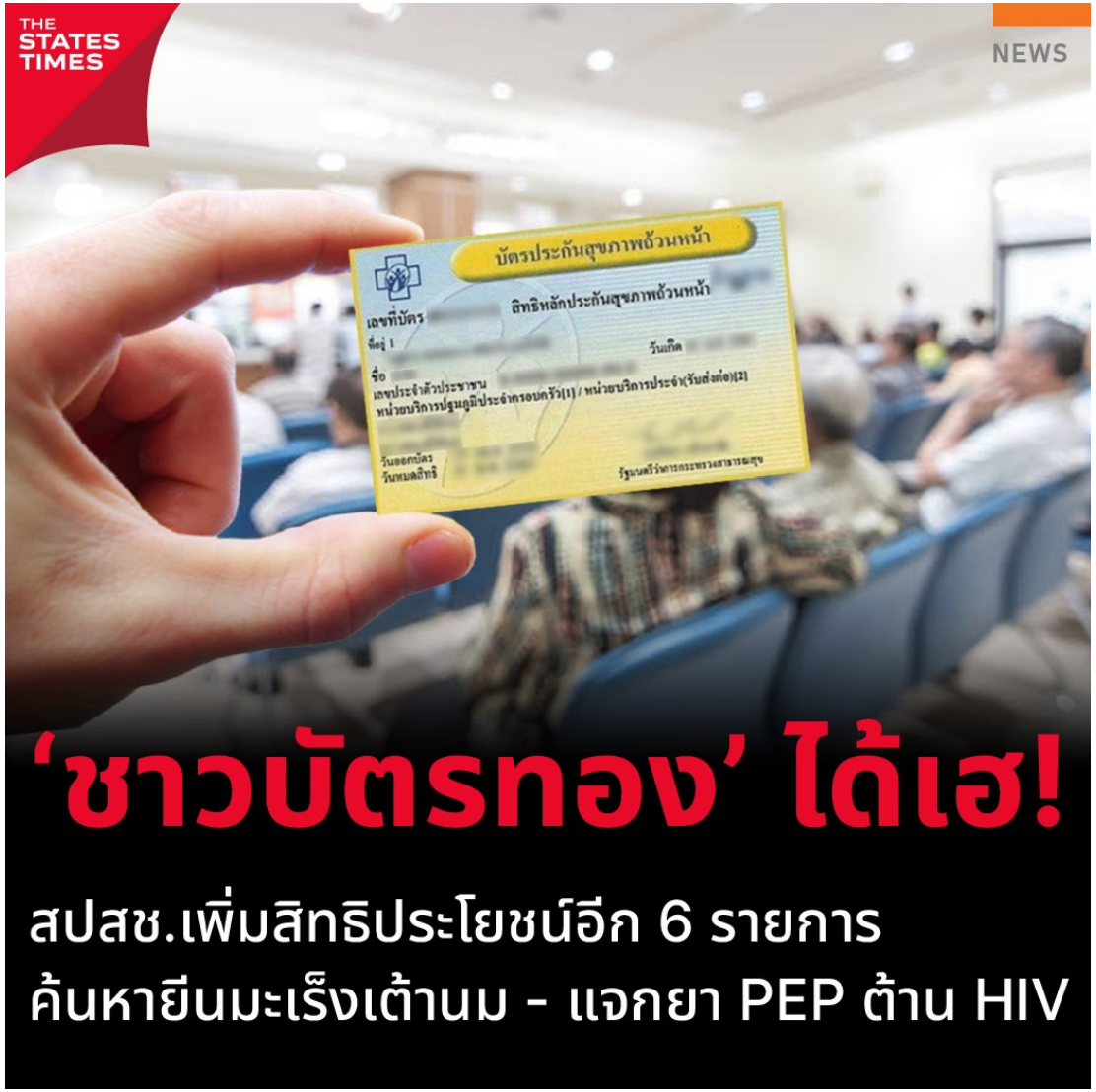
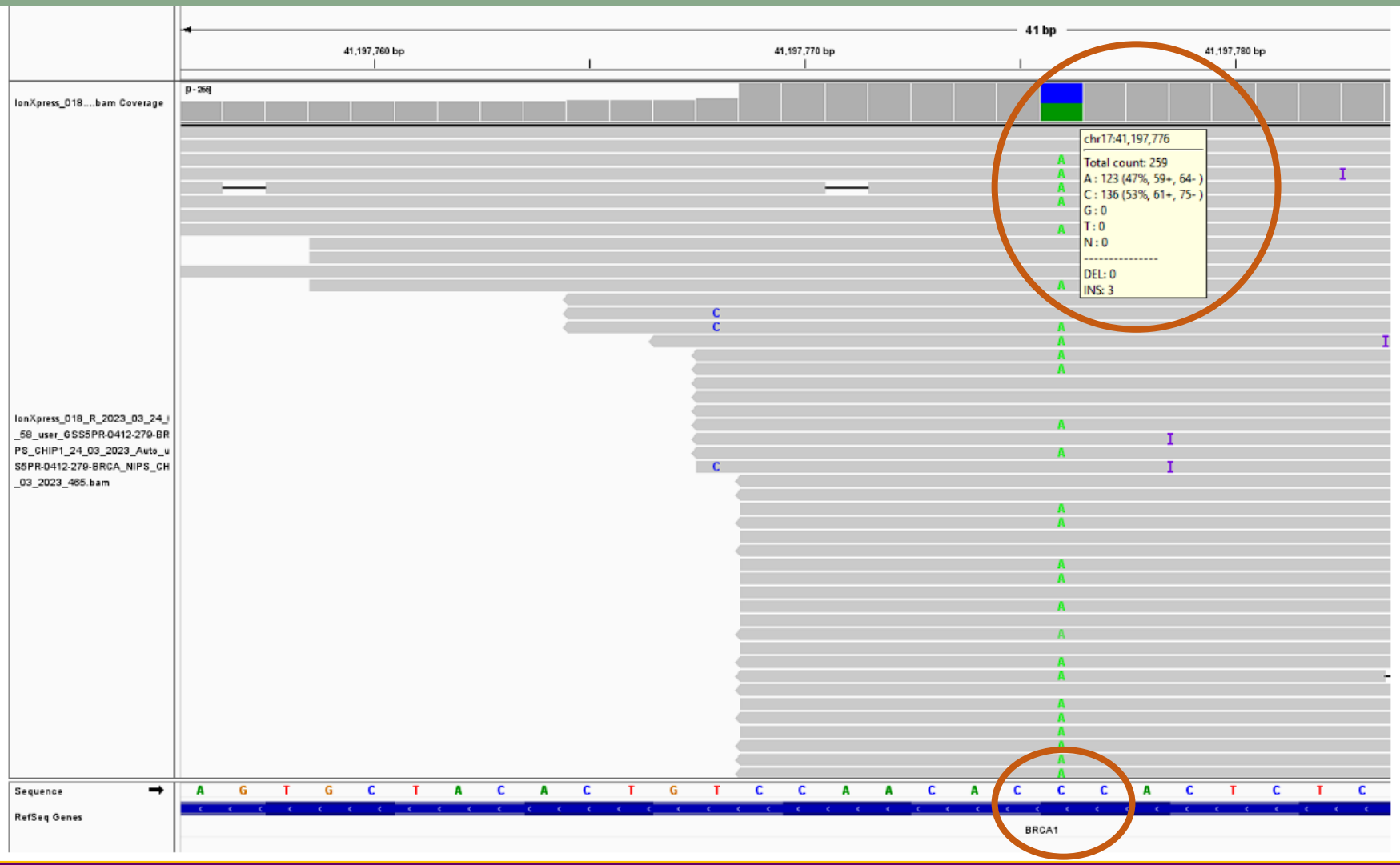
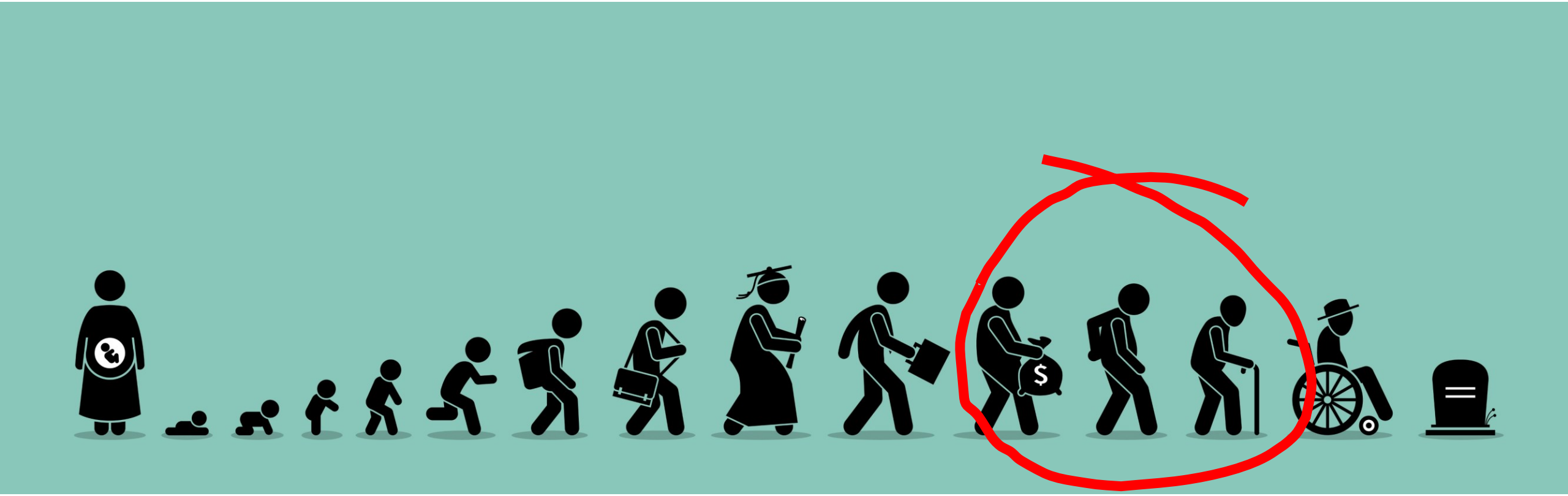

THE STATES TIMES
NEWS
บัตรประกันสุขภาพถ้วนหน้า
สิทธิหลักประกันสุขภาพถ้วนหน้า
‘ชาวบัตรทอง’ ได้เฮ!
สปสช.เพิ่มสิทธิประโยชน์อีก 6 รายการ
ค้นหายีนมะเร็งเต้านม - แจกยา PEP ต้าน HIV

# Targeted sequencing : BRCA1&BRCA2





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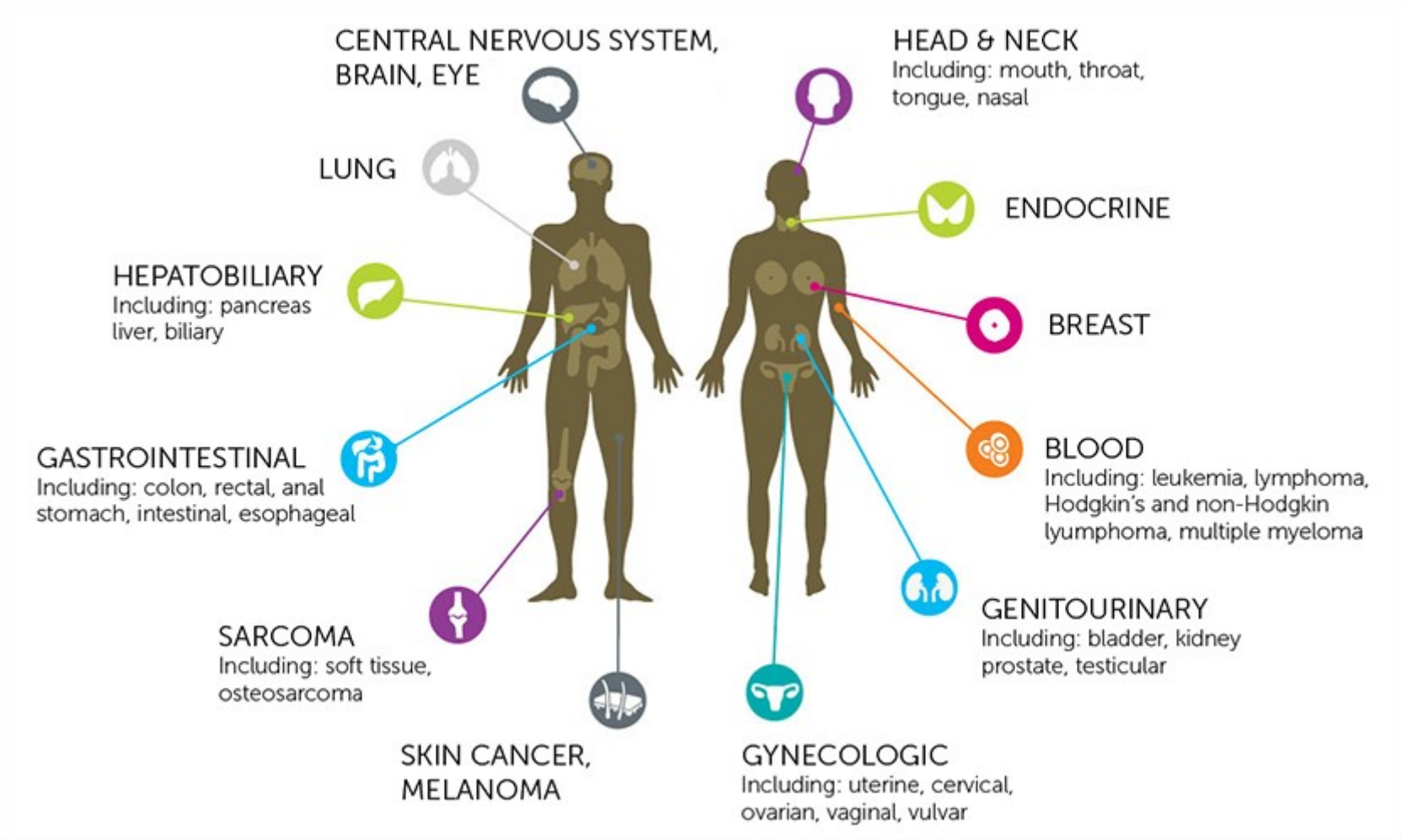
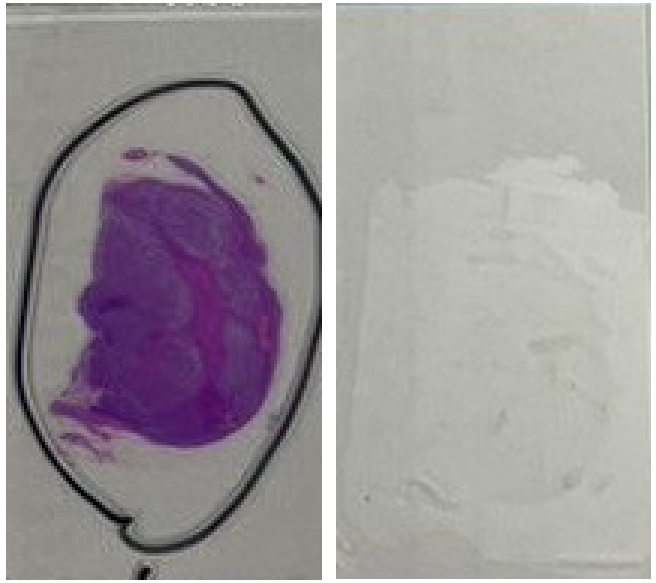
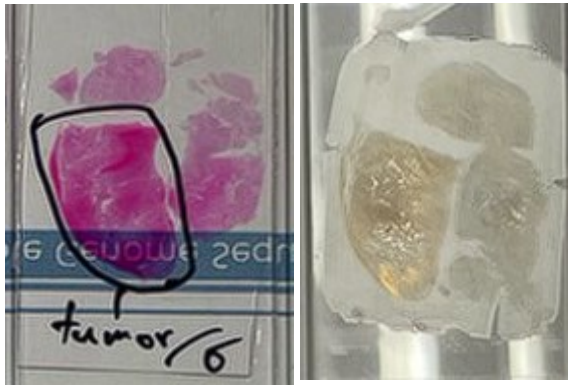



https://breastin.info/

https://medsurgeindia.com/

# Targeted / WES sequencing : Fresh/FFPE tissue





Example Labs
123 Street
City, State 00000
Tel +1 000-000-0000
email@example.com
www.example.com

Case Number: [redacted]



## Patient Information

**Patient Name:** [redacted]
**Date of Birth:** -
**Physician:** [redacted]
**Diagnosis:** -

**HN:** [redacted]
**Gender:** Female
**Hospital:** [redacted]

## Sample Information

**Sample ID:** -
**Collection Date:** 25 August 2022
**Limits of Detection %** 0.1069 - 0.1474

**Sample Type:** Blood
**Received Date:** 30 August 2022

## Sample Cancer Type: Non-Small Cell Lung Cancer

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**Report Highlights**
4 Relevant Biomarkers
16 Therapies Available
235 Clinical Trials

## Relevant Non-Small Cell Lung Cancer Findings

| Gene | Finding | Gene | Finding |
|---|---|---|---|
| ALK | None detected | MET | None detected |
| BRAF | ***BRAF wild type*** | NTRK1 | None detected |
| EGFR | ***EGFR exon 19 deletion, EGFR A750P*** | NTRK3 | None detected |
| ERBB2 | None detected | RET | None detected |
| KRAS | None detected | ROS1 | None detected |

## Relevant Biomarkers

| Genomic Alteration | Relevant Therapies (In this cancer type) | Relevant Therapies (In other cancer type) | Clinical Trials |
|---|---|---|---|
| *EGFR exon 19 deletion*<br>epidermal growth factor receptor<br>Allele Frequency: 55.99% | **afatinib** [1,2]<br>**bevacizumab* + erlotinib** [2]<br>**dacomitinib** [1,2]<br>**erlotinib** [1,2] | None | 222 |

**Public data sources included in relevant therapies:** FDA[1], NCCN, EMA[2], ESMO
*** Includes biosimilars**

Optional left-aligned text block, one line or multiple lines. Lines will span full width of page width and then wrap to new line within footer area.

**Disclaimer:** The data presented here is from a curated knowledgebase of publicly available information, but may not be exhaustive. The data version is 2022.10(005).The content of this report has not been evaluated or approved by FDA, EMA or other regulatory agencies.



## Relevant Biomarkers (continued)

| Genomic Alteration | Relevant Therapies (In this cancer type) | Relevant Therapies (In other cancer type) | Clinical Trials |
|---|---|---|---|
| | **erlotinib + ramucirumab** [1,2]<br>**gefitinib*** [1,2]<br>**osimertinib** [1,2]<br>atezolizumab + bevacizumab + chemotherapy<br>bevacizumab + gefitinib<br>gefitinib + chemotherapy<br>osimertinib + chemotherapy | | |
| *EGFR A750P*<br>epidermal growth factor receptor<br>Allele Frequency: 56.01% | None | None | 92 |
| *FGFR2 amplification*<br>fibroblast growth factor receptor 2 | None | None | 15 |

**Public data sources included in relevant therapies:** FDA[1], NCCN, EMA[2], ESMO
*** Includes biosimilars**

**Alerts informed by public data sources:** Contraindicated, Resistance, Breakthrough, Fast Track

| | |
|---|---|
| *EGFR exon 19 deletion* | **patritumab deruxtecan** [1]<br>**osimertinib + quaratusugene ozeplasmid** [1] |
| *BRAF wild type* | **dabrafenib** [1,2] |
| *EGFR A750P* | **osimertinib + quaratusugene ozeplasmid** [1] |
| *FGFR2 amplification* | **Debio 1347** [1] |

**Public data sources included in alerts:** FDA[1], NCCN, EMA[2], ESMO

## Variant Details

### DNA Sequence Variants

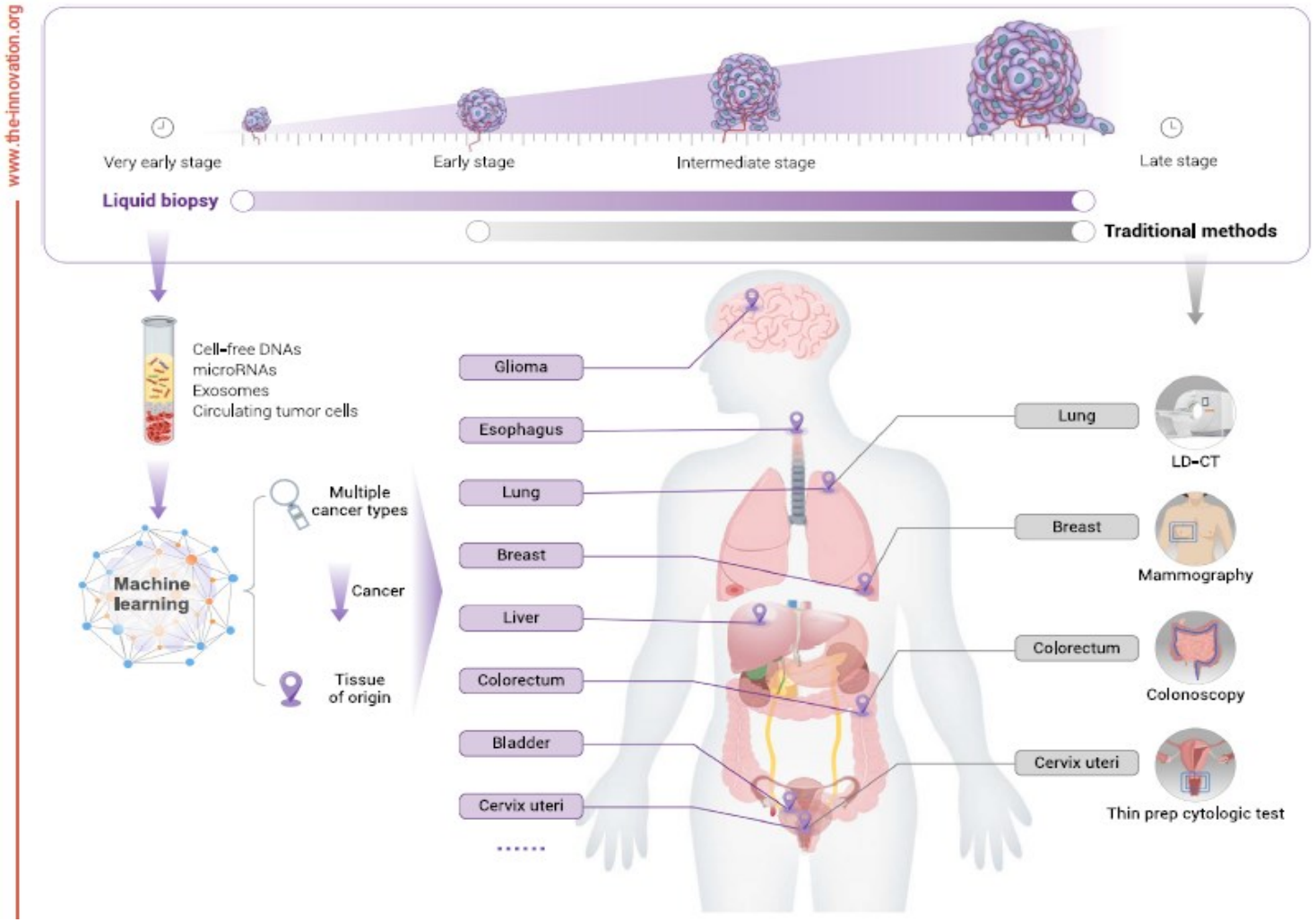
| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect |
|---|---|---|---|---|---|---|---|
| EGFR | p.(L747_A750delinsP) | c.2239_2248delTTAAGAGAAGinsC | p.L747_A750P | chr7:55242469 | 55.99% | NM_005228.5 | nonframeshift Block Substitution |
| EGFR | p.(A750P) | c.2248G>C | p.A750P | chr7:55242478 | 56.01% | NM_005228.5 | missense |

### Copy Number Variations

| Gene | Locus | Copy Number |
|---|---|---|
| FGFR2 | chr10:123245035 | 3.12 |

**Disclaimer:** The data presented here is from a curated knowledgebase of publicly available information, but may not be exhaustive. The data version is 2022.10(005).

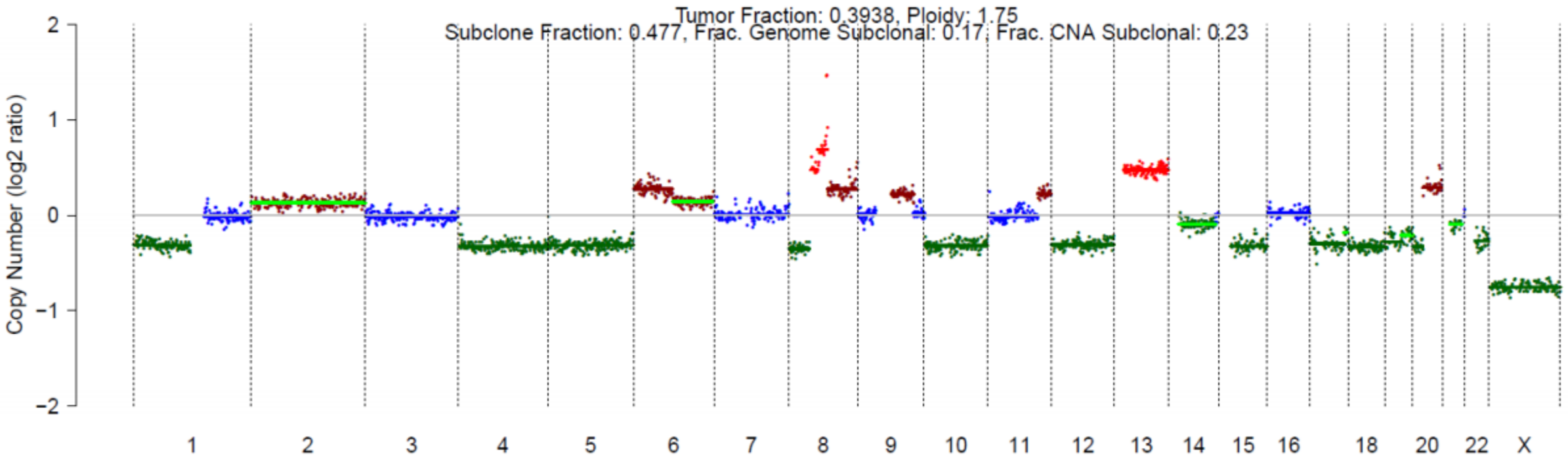
# Liquid biopsy : ctDNA in blood



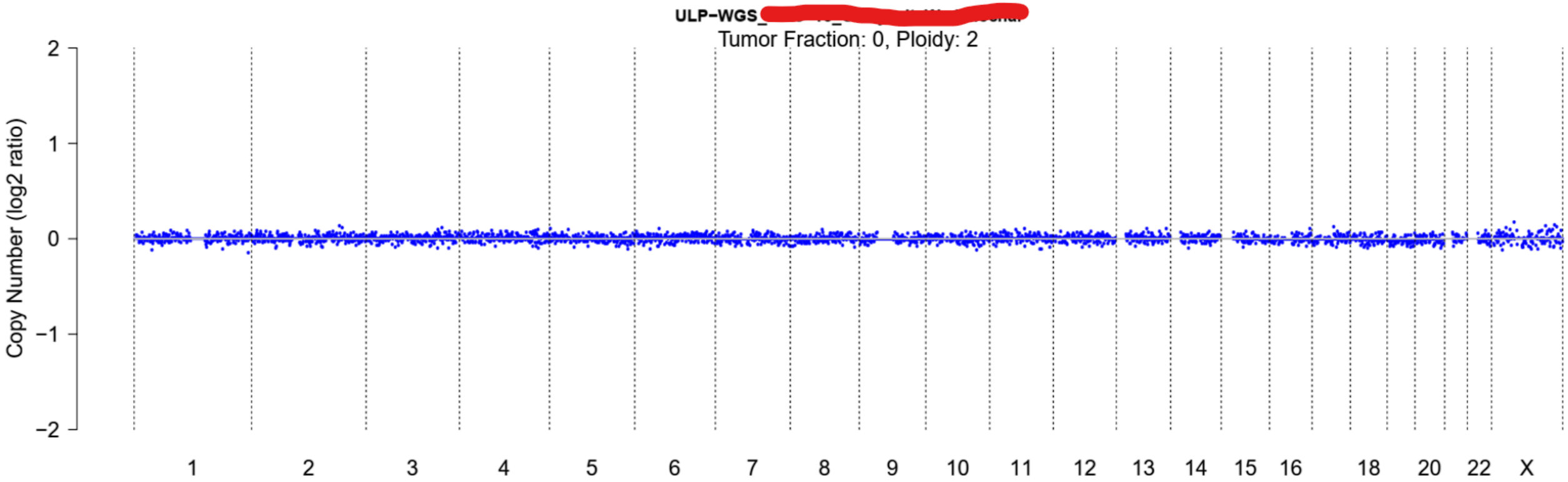
The 52-gene panel includes:

- Hotspot genes (SNVs) and short indels: AKT1, ALK, AR, ARAF, BRAF, CHEK2, CTNNB1, DDR2,EGFR, ERBB2, ERBB3, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, NTRK1, NTRK3, PDGFRA, PIK3CA, RAF1, RET, ROS1, SF3B1, SMAD4, SMO
- Gene fusions: ALK, BRAF, ERG, ETV1, FGFR1, FGFR2, FGFR3, MET, NTRK1, NTRK3, RET, ROS1
- MET exon 14 skipping
- Copy number genes (CNVs): CCND1, CCND2, CCND3, CDK4, CDK6, EGFR, ERBB2, FGFR1, FGFR2, FGFR3, MET, MYC
- Tumor suppressor genes: APC, FBXW7, PTEN, TP53

Targeted gene panel : SNV, indels, gene fusion, CNV, whole gene




CNA from plasma of a patient with solid tumor, stage IV




Normal CNA profile

# Future perspective

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## 4 New Technologies

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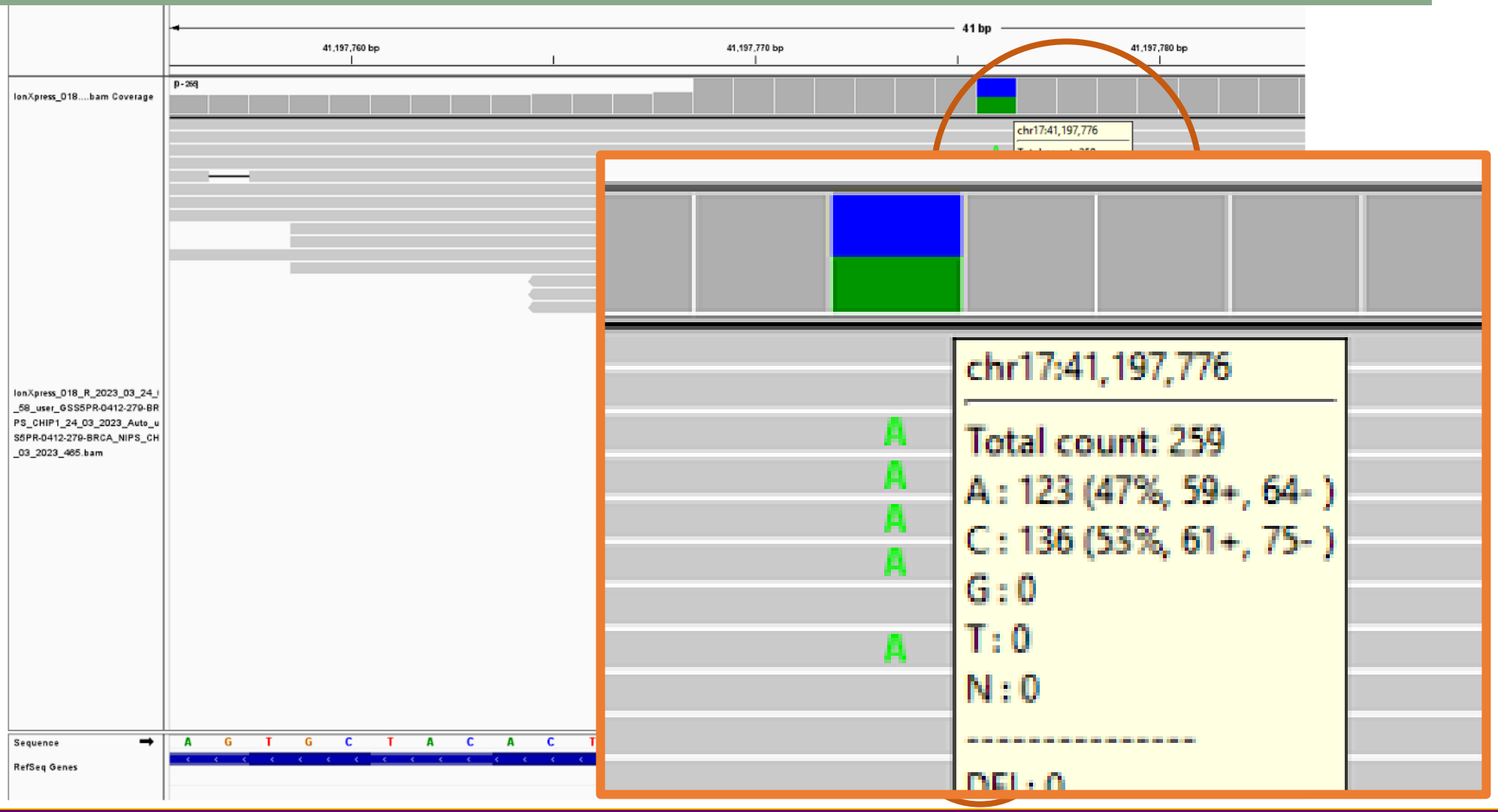
**New Technologies in Development**

https://revolugen.co.uk/market-applications/dna-sequencing.html

# Targeted sequencing : BRCA1&BRCA2



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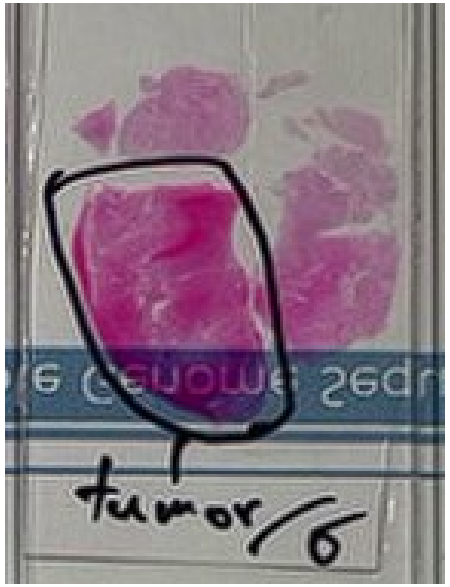
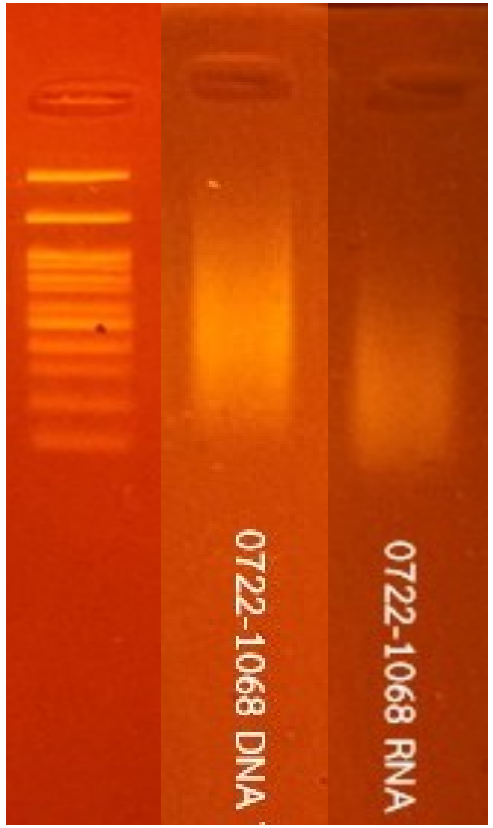

tumor/6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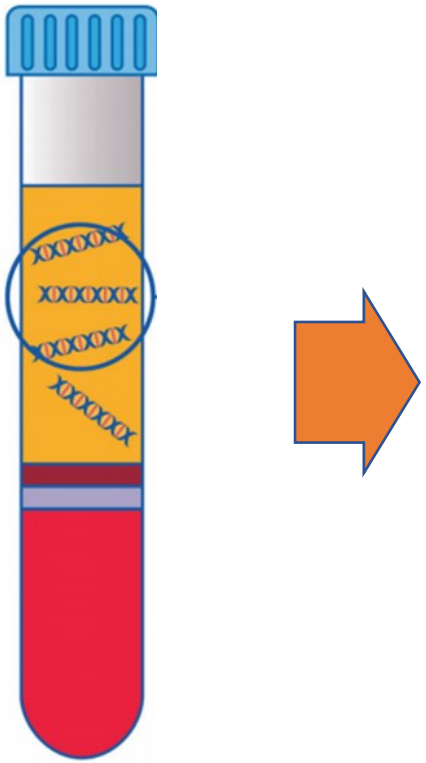
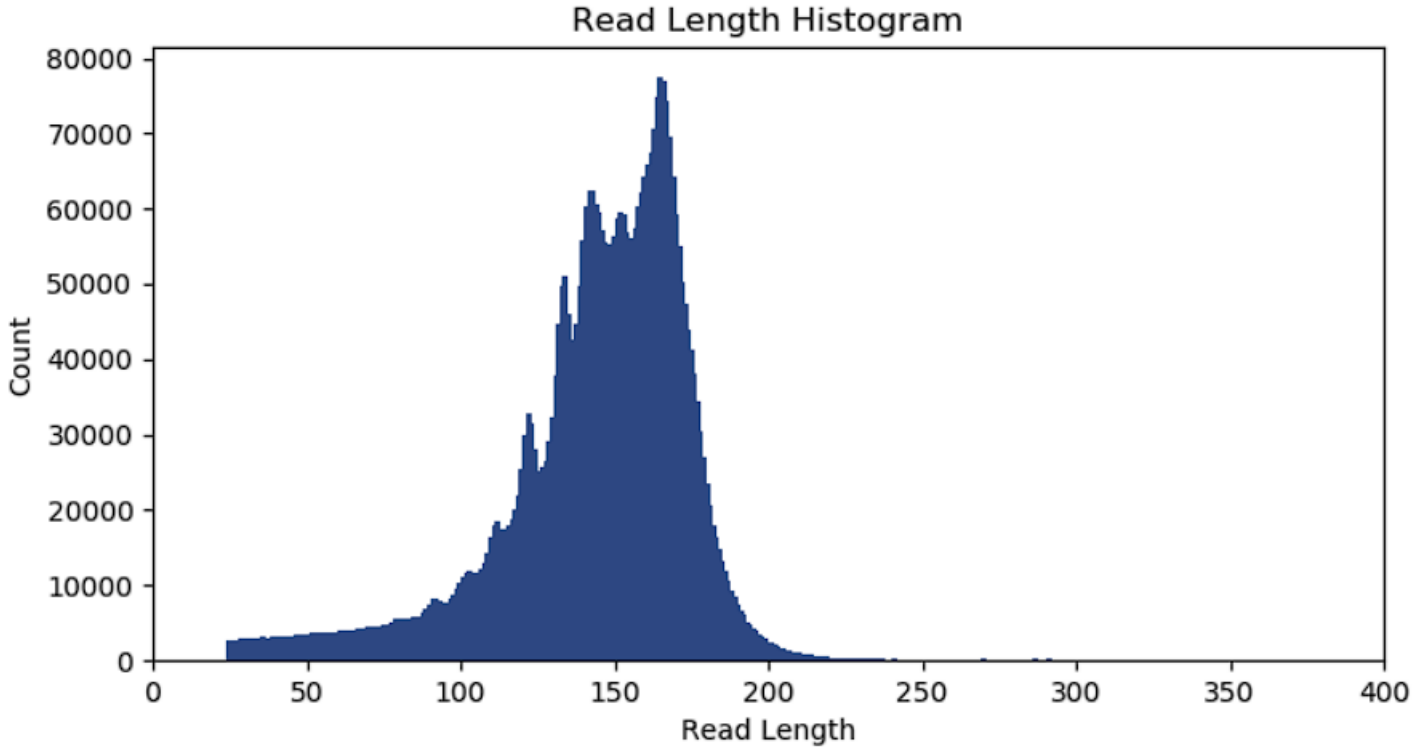
0722-1068 DNA




Read Length Histogram
Count
80000
70000
60000
50000
40000
30000
20000
10000
0
0
50
100
150
200
250
300
350
400
Read Length

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# Thank you

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# Update State of the Art Genomic Platforms

# ช่วงถามตอบ

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