Advancing Personalized Medicine Through Genomic Testing

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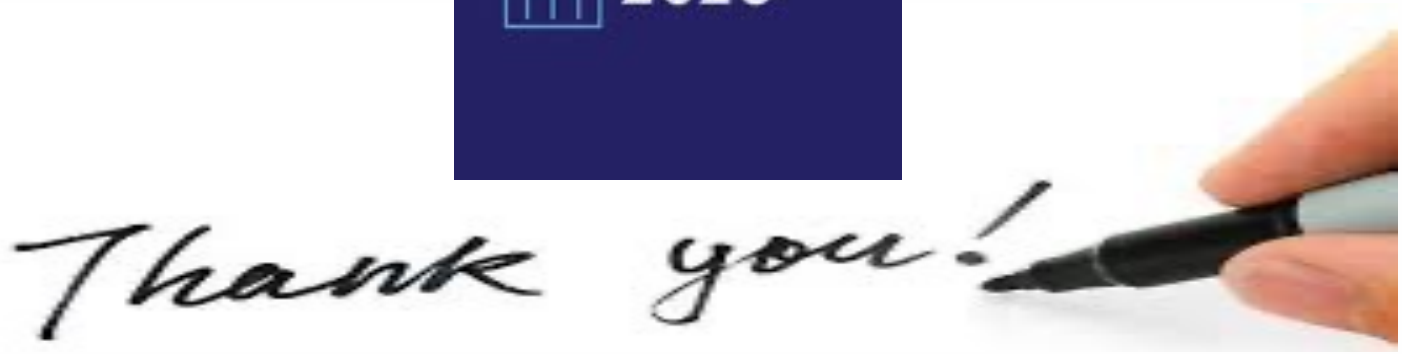

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Today's Lecture

Brief introduction to MedLabs

Technical Introduction to Genomics

Genomic Testing Capabilities

The Current and Future of Genomic Testing

Conclusion

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GENOMICS
By MedLabs

An Introduction to MedLabs

Established in **1993** as a Jordanian Private Shareholding Company

MedLabs is the one of the **largest** medical laboratory networks in the Middle East

72 labs in Jordan, Palestine, Iraq and Kurdistan, UAE and Ireland

~250 laboratories in the Middle East and Gulf Countries refer advanced tests to Medlabs referral laboratory.

Over 680 employees including 16 MDs, 8 PhDs, 17 MScs & MBAs

We are a non-commission based company!

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Celebrating 33 years!

CAP Accredited for all tests performed within the reference lab since 2011

ISO15189 Accredited since 2009

ISO9001 Accredited since 2007

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Technical Introduction to Genomics

- Humans have around 50 Trillion cells, each contain tightly packed 23 pair of chromosomes (50% from mother and 50% from father).
- DNA is made up of 4 basic letters (nucleotides) A C G T –DNA Alphabet
- 3.2billion letters that make each complete genome are equivalent to ca. 1800 books each with 500 pages.
- We only understand 2-3% of the genome, around 22.000 genes with mutations causing around 15,000 different diseases.
- We inherit mutations in the code or typos giving rise to disease (mutations)
- Diseases can occur in different ages (recessive in childhood and dominant in adulthood)
- Genetics is just one piece of the puzzle: environment, nutrition, lifestyle, age, infection...etc., are as important

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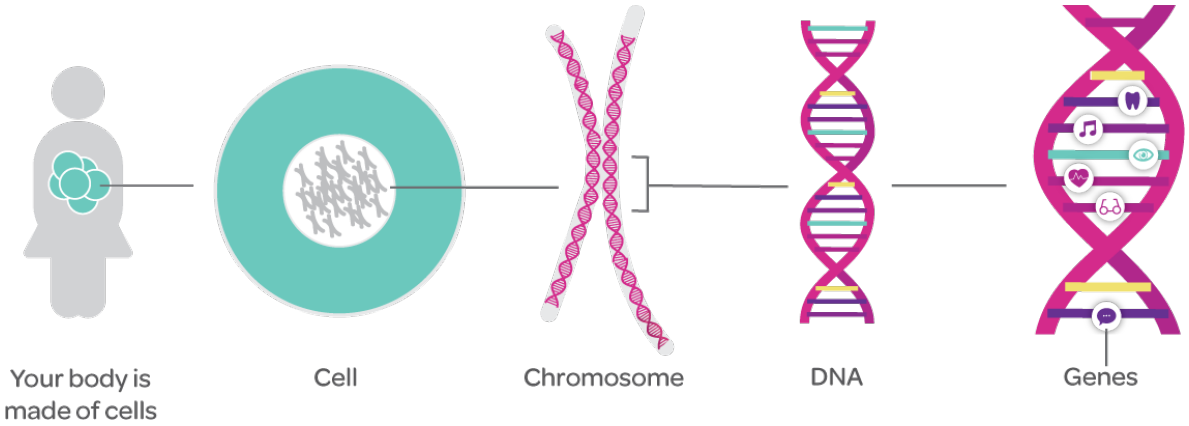
Technical Introduction to Genomics



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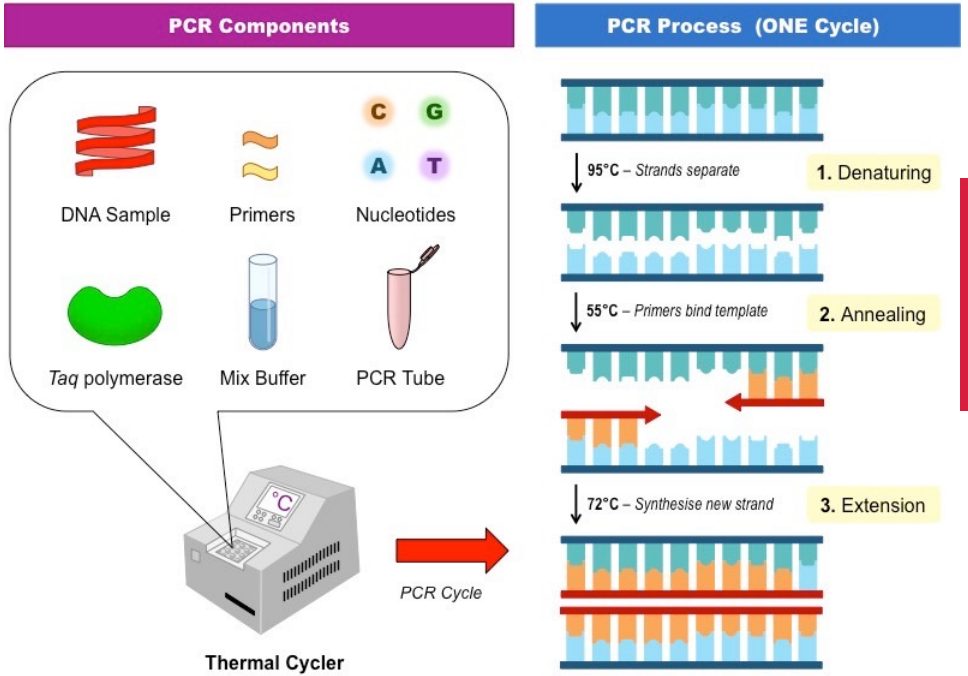
Technical Introduction to Genomics



PCR the backbone of genetics

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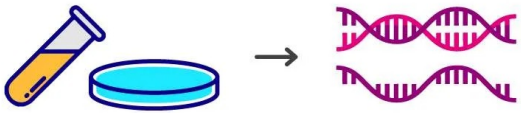
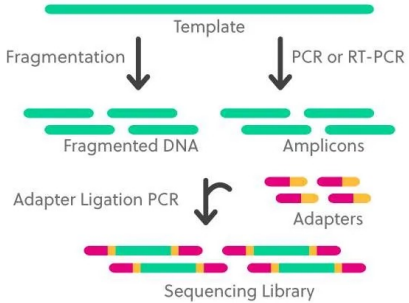
Technical Introduction to Genomics

STEP 1:
Extraction



STEP 2:
Library Prep



STEP 3:
Sequencing

STEP 4:
Analysis

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Technical Introduction to Genomics

- What are we producing?
- Why are there differences in price?
- WGS vs WES vs Targeted panels

Test	Genes	Depth	Coverage	Information content	Likelihood of detecting a specific mutation
Whole Genome Sequencing	~20.000	~30x	~99% >10x	~3.2GB	~97.6%
Whole Exome Sequencing	~20.000	~100x	~97% >10x	~60MB	~93.2%
NGS Panel	10-1000	~200x	~98% >20x	20-400KB	~96.1%
Single Gene Sequencing (Sanger)	1	NA	NA	0.2-5KB	~99.9%

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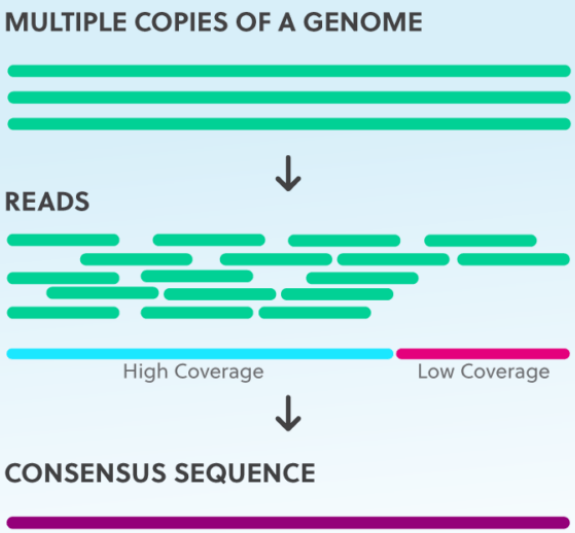
Technical Introduction to Genomics

Always ask for the technical information, not just about accreditations



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Genomic Testing Capabilities

Direct to consumer

Clinical genetic testing

Multiple Genes = Each With Small Effect

One Gene = Big Effect

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Genomic Testing Capabilities (Lifestyle)

Multiple Genes = Each With Small Effect

Saccharomyces cerevisiae
Listeria monocytogenes
Enterococcus faecalis
Escherichia coli
Staphylococcus aureus
Bacillus subtilis
Salmonella enterica
Pseudomonas aeruginosa
Lactobacillus fermentum

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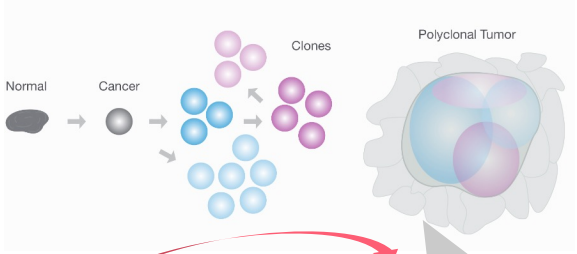
Genomic Testing Capabilities

Hereditary cancer

Non hereditary cancer



Clonal evolution

Tumor and unaffected cells, have the causative mutation

e. BRCA1/2, TP53

Somatic mutations of the tumor tissue

A tumor is a mix of normal and tumor genomes

Advanced polyclonal tumors: several clones

- Unique mutations
- Individual treatment response
- Possible relapse after treatment, evolution of a founding clone, survival of a subclone

10%

90%

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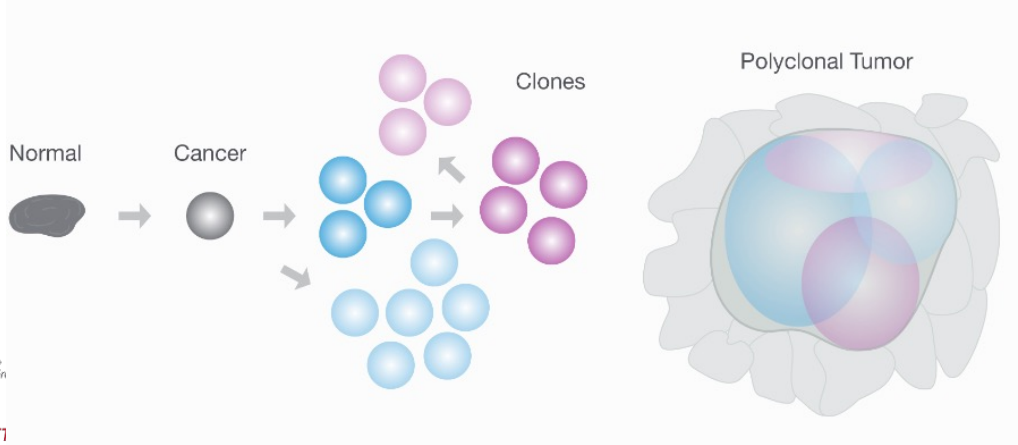
Genomic Testing Capabilities

Tailors' cancer care: For cancer patients, testing tumour DNA helps doctors pick specific, targeted therapies that work best for that exact type of cancer

Avoids bad side effects: It shows which medicines match your body, helping you avoid drugs that will not work or might harm you



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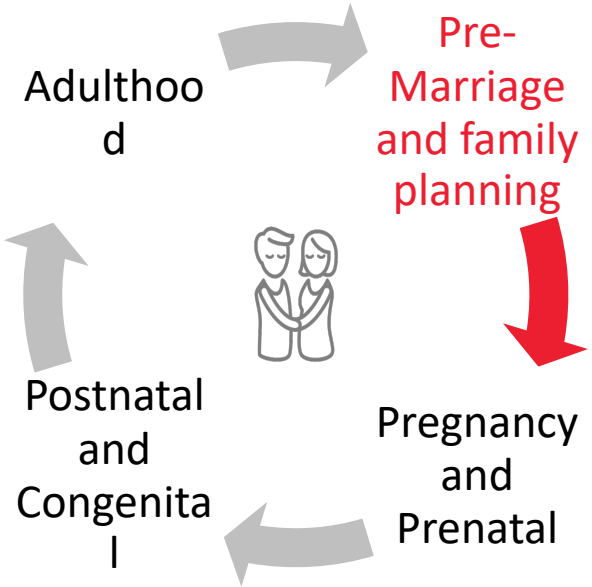
Genomic Testing Capabilities

And then the list goes on

- Rare diseases
- Inherited diseases
- IVF
- NIPT



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The Current and Future of Genomic Testing

- Cost of testing
- Availability and TAT
- Education amongst physicians
- Country wide genome projects to understand the population and prepare for the future
- More sharing of data means a better outcome for all (VUS)
- A powerful tool that needs REGULATION

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Conclusion

- The importance of Accreditation can not be understated
- Who owns the data
- Health insurance ethics
- AI will start to revolutionise the speed and outcome of data
- Remove the stigma of shame and talk about consanguinity
- Public/Private cooperation

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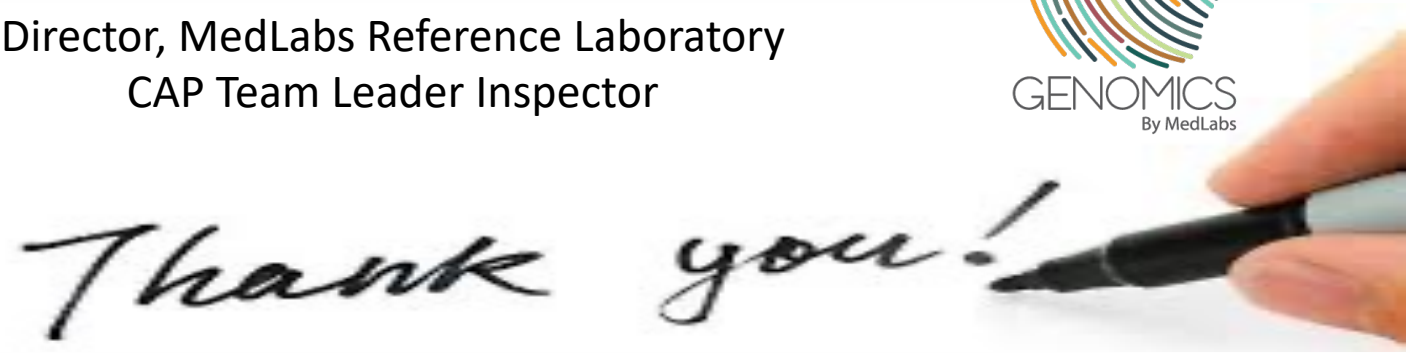

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