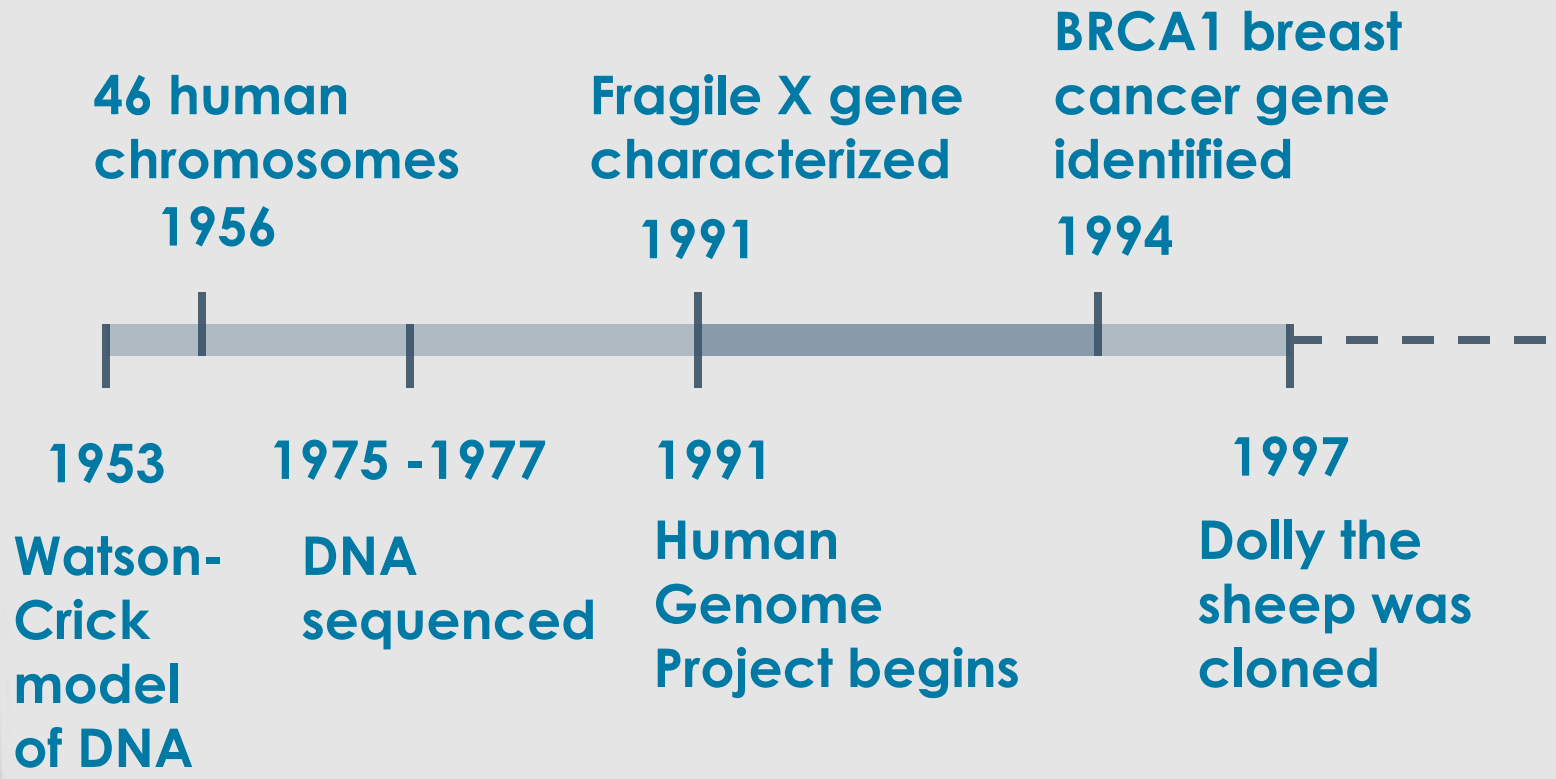


Molecular & Cytogenetic Testing



Molecular Level

FISH, PCR &
gene analysis

Forensic

STDs

- Chlamydia
- Neisseria
- HPV
- HCV
- HSV

Prenatal

- α -Thalassemia
- Blood group
- Cystic Fibrosis

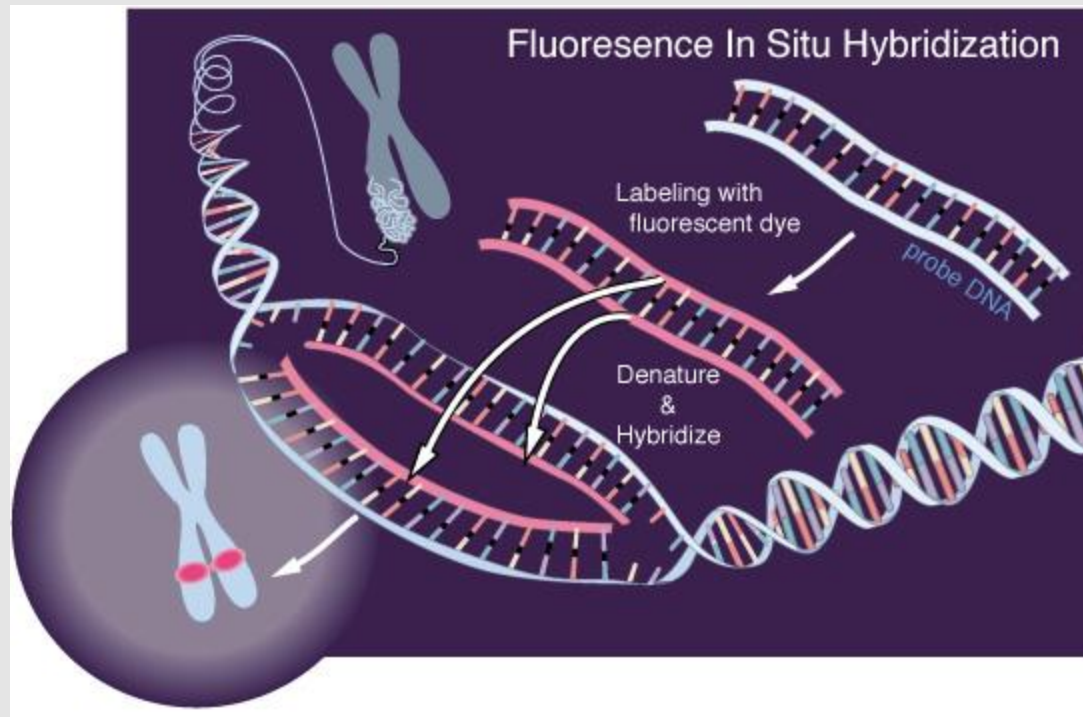
Diagnosis

- Y-chromosome microdeletion

Carrier

- Factor V
- MTHFR
- Factor II
- Muscular dystrophy

Fluorescence In Situ Hybridization (FISH)



Examples of FISH tests available at MedLabs



Philadelphia Chromosome BCR/abl
Trisomy 13 (Patau's Syndrome)
Trisomy 18 (Edward's Syndrome)
Trisomy 21 (Down's Syndrome)
X & Y

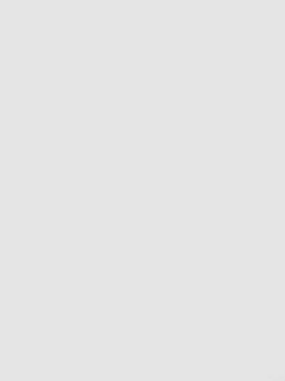
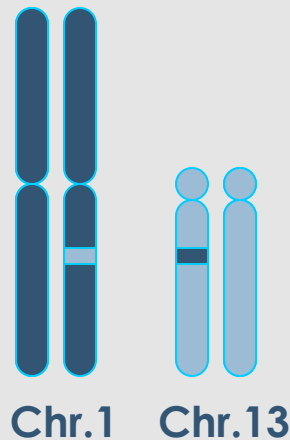
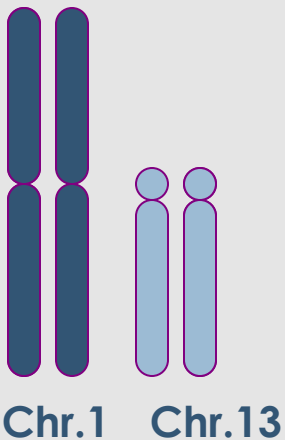
PGD case



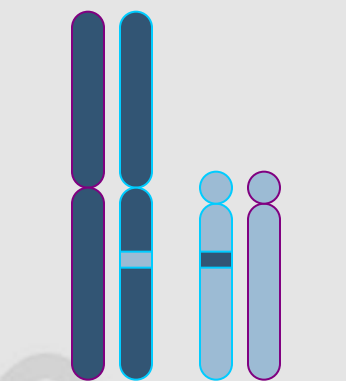
○ Sahar

46,XY,t(1;13)(q32;q14)

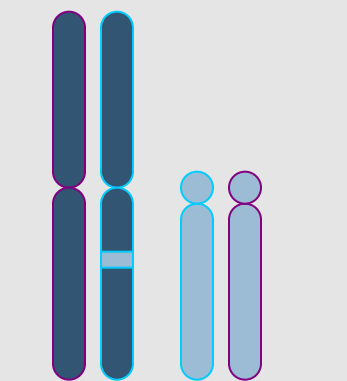
Fares



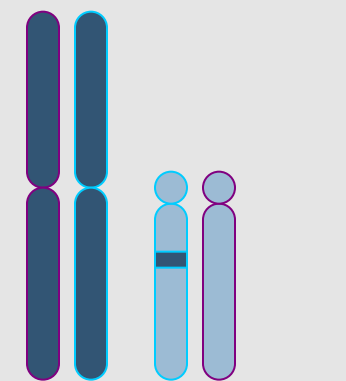
Normal



Balanced translocation (like dad)

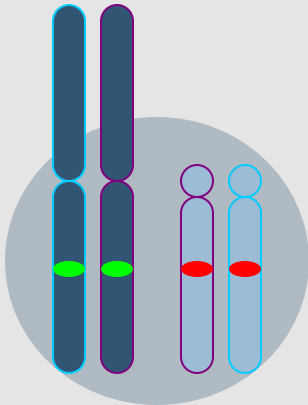
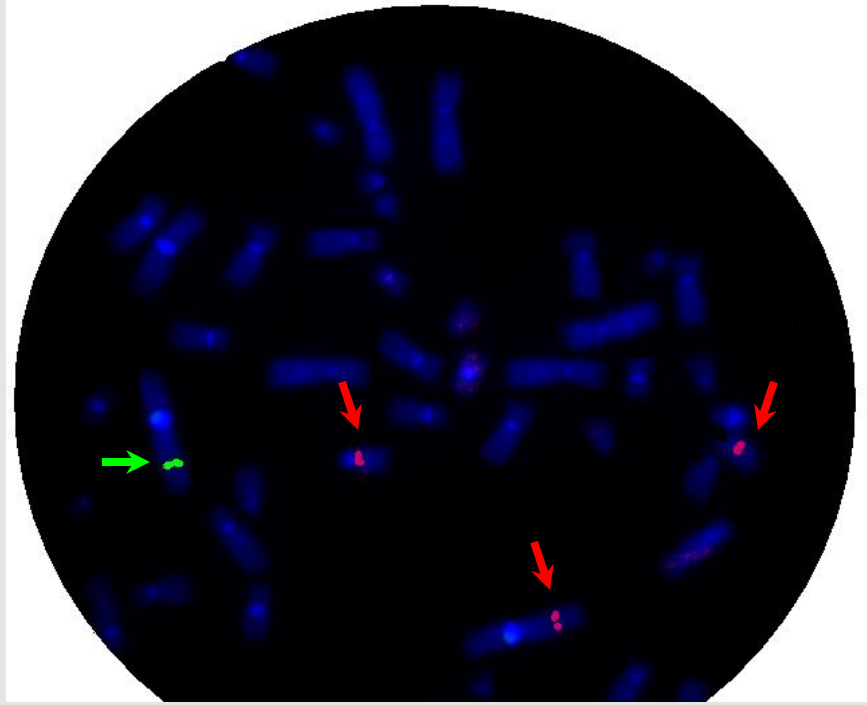
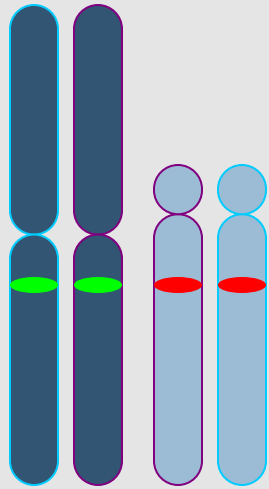


Partial - Tri 13
Mon 1

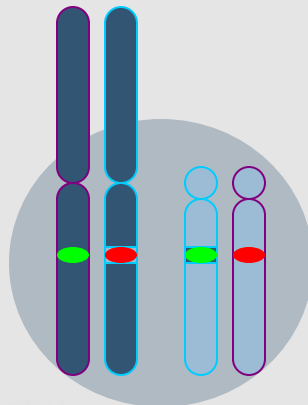


Partial - Tri 1
Mono 13

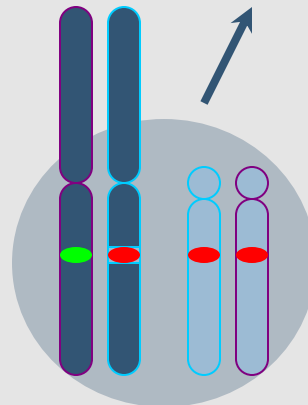
PGD case



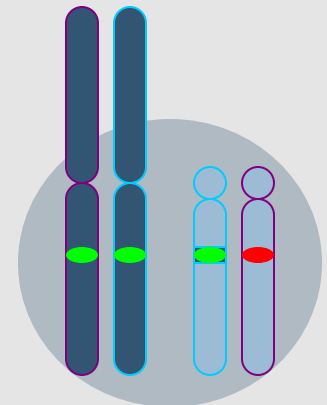
Normal



Balanced translocation
(like dad)



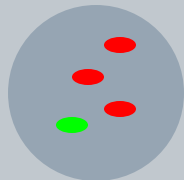
Unbalanced translocation



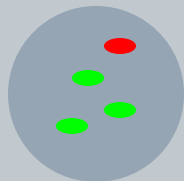
Unbalanced translocation

PGD case

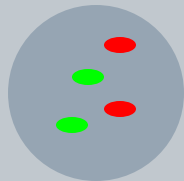
Attempt # 1




3 embryos
Tri 13, mono 1



1 embryo
Tri 1, mono 13

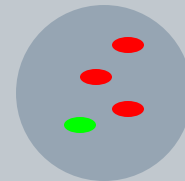


1 embryo
Normal / carrier

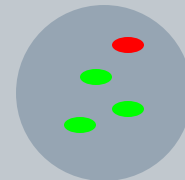
Gender identification →  XX

Embryo transferred but
conception failed

Attempt # 2



1 embryo
Tri 13, mono 1



3 embryos
Tri 1, mono 13



1 embryo
No signal



2 embryos
Normal / carrier

Delivered a normal baby
girl

Examples of PCR tests available at MedLabs



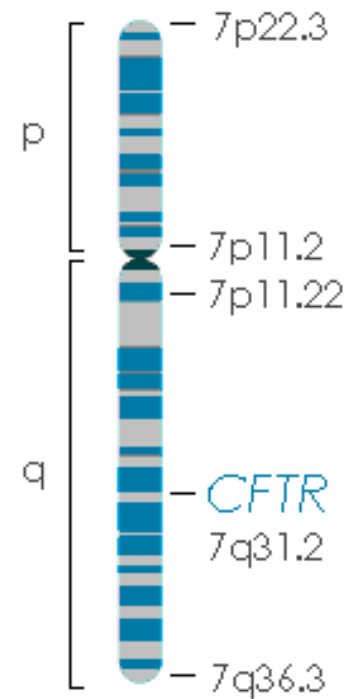
Activated Protein C Resistance
Beta-Globin gene (β -Thal.)
CVD
Chlamydia T.
Cystic Fibrosis (36 Mutations)
CMV
FMF
Factor II Prothrombin A Mutation
Gonococcal
H. Pylori (DNA)
HBV (DNA)
HCV (RNA)
HCV Genotyping (RT-PCR)
Hereditary Haemochromatosis
Herpes Simplex (DNA)

HLA -ABC
HLA-B27
HLA-B5
HLA-DR
Human Papiloma Virus Genotyping
H1N1
MTHFR
Mycobacterium Tuberculosis (DNA)
Mycoplasma Pneumonia
Neisseria Gonorrhoea
Parrovirus-B19
Prothrombin G20210A
Sickle Cell gene mutation
Thrombophilia Genetic Panel
Y-Chromosome
Alpha-Thalassemia

Cystic Fibrosis

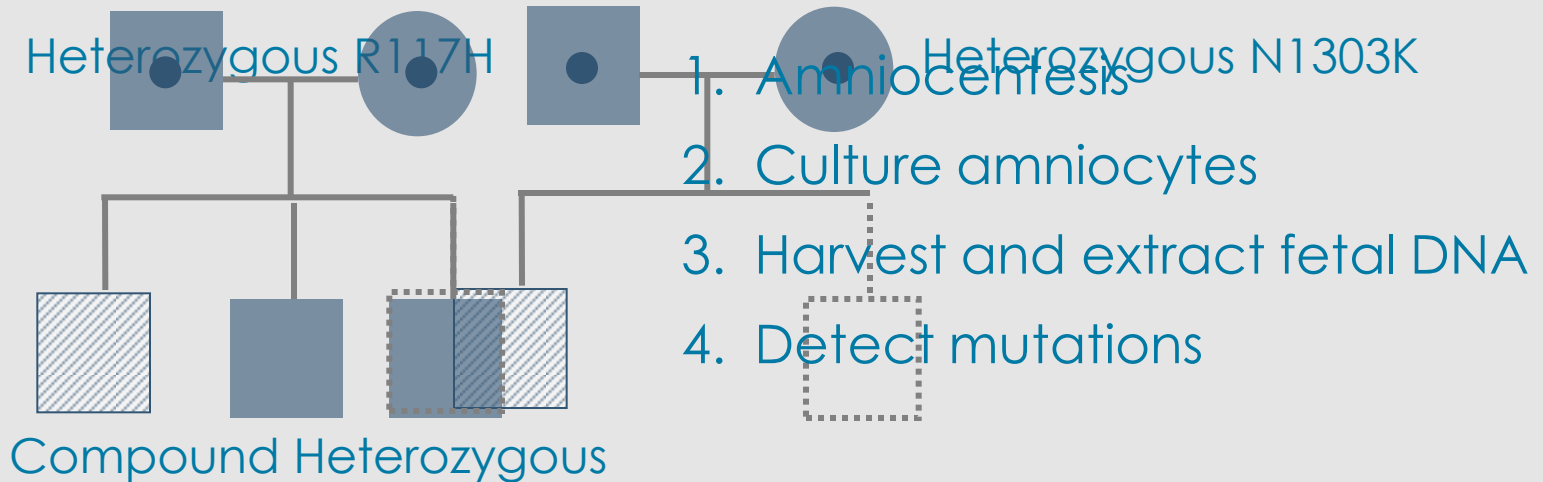
- Prevalence of the CFTR mutation in the general population → 1:25
- Most common autosomal recessive disease among Caucasians, affecting 1 in 2500 live births
- 900 different mutations have been identified
- The most frequent → $\Delta F508$

Chromosome 7



CFTR case

- Prenatal Genetic Diagnosis



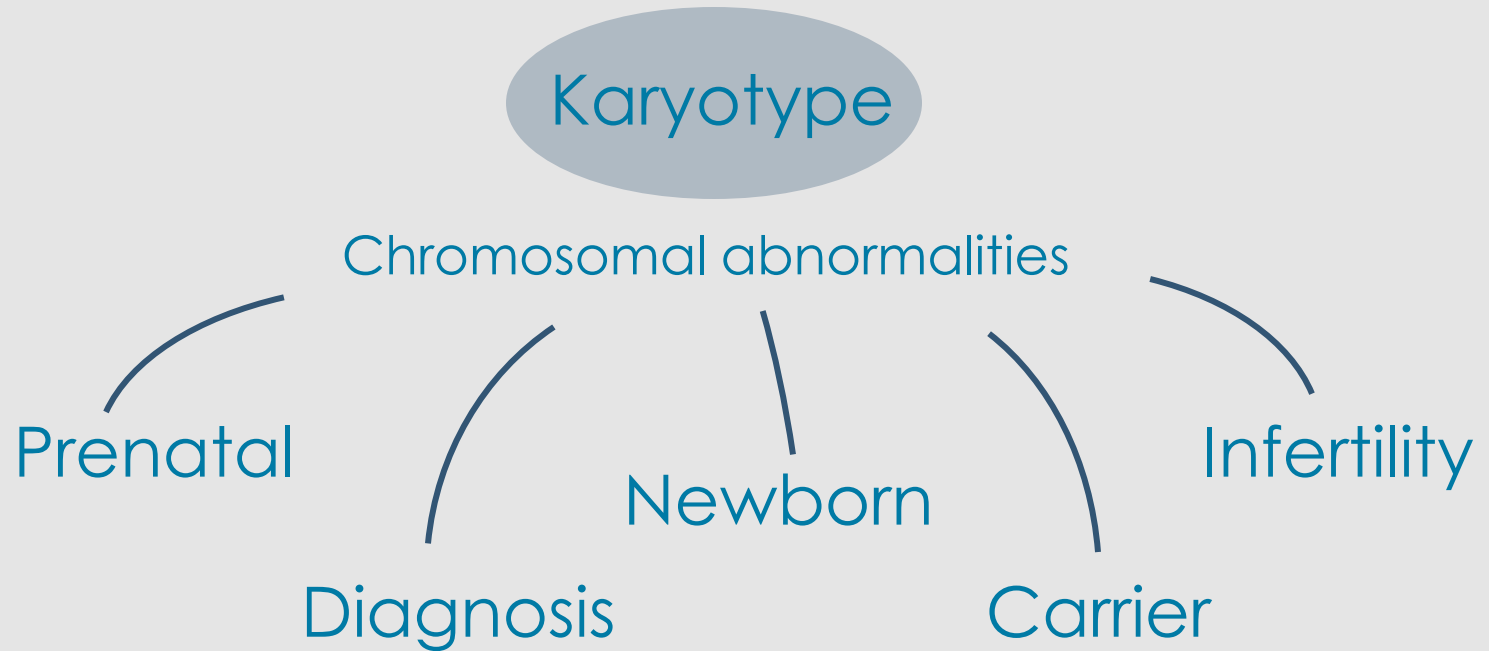
- To ensure that there was no maternal DNA contamination, amniocytes were also karyotyped
- Similar procedure is followed for β -thalassemia and sickle cell anemia

AmnioPCR

QF-PCR: **Q**uantitative **F**luorescence
Polymerase **C**hain **R**eaction

- Used for the detection of aneuploidy of whole chromosomes (13, 18, 21, X and Y) in amniotic fluid samples
- STR regions of DNA extracted from uncultured amniotic fluid fetal cells are amplified by PCR

Karyotype analysis



- Down syndrome
- Klinefelter
- Fragile X

- Translocations
- Deletions
- Duplications

Sexually Transmitted Diseases & Reproduction



Human papilloma virus genotyping

Herpes simplex I and II detection

Hepatitis B viral load and genotyping

Hepatitis C viral load and genotyping

Chlamydia and Neisseria detection

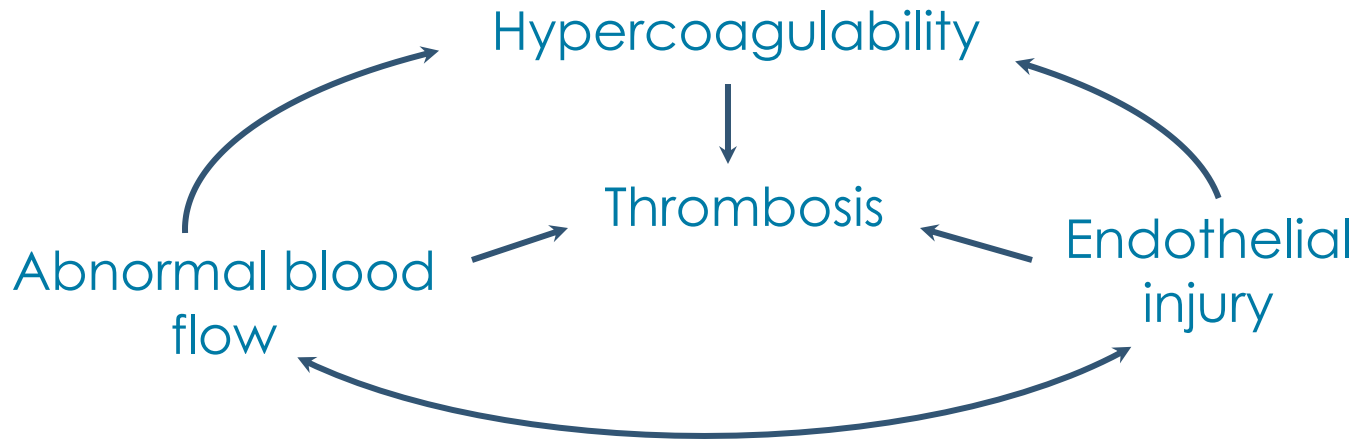
HIV viral load

A large, faint grey line graph is overlaid on the slide. It starts with a very high, sharp peak on the left side, then drops to a low baseline with several smaller, rounded peaks and troughs extending across the bottom of the slide.

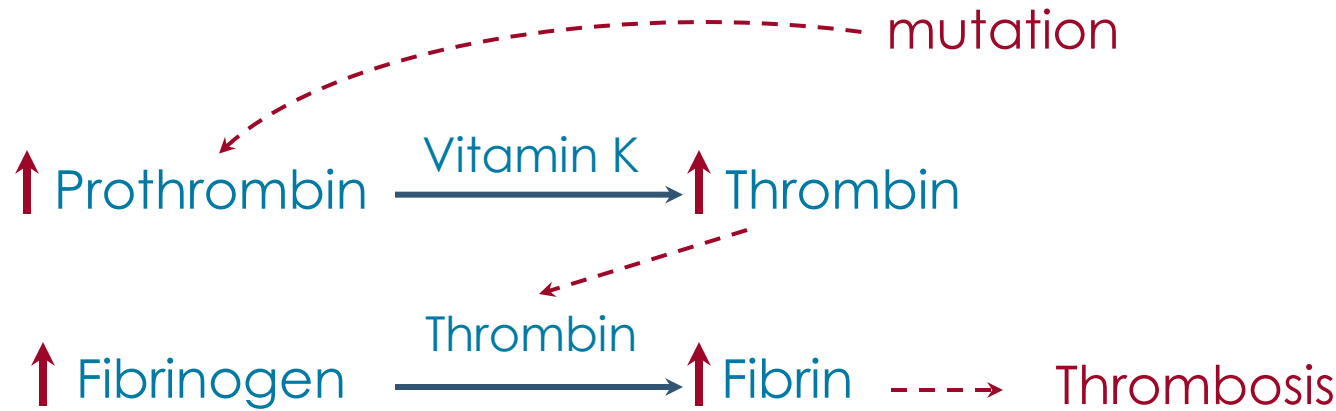
Thank you



Factor V Leiden (Activated Protein C Resistance)



Factor II (Prothrombin G20210A)





Genetic tests in male infertility

- Karyotype
- Y chromosome microdeletions
- Cystic Fibrosis

Lower incidence

- Aneuploidy in spermatozoa by FISH
- KALI gene for Kallmann syndrome
- Androgen receptor
- 5 alpha reductase



Genetic tests in female infertility

- Karyotype
- Fragile X
- Cystic Fibrosis

Rare incidence

- KAL1 gene for Kallmann syndrome