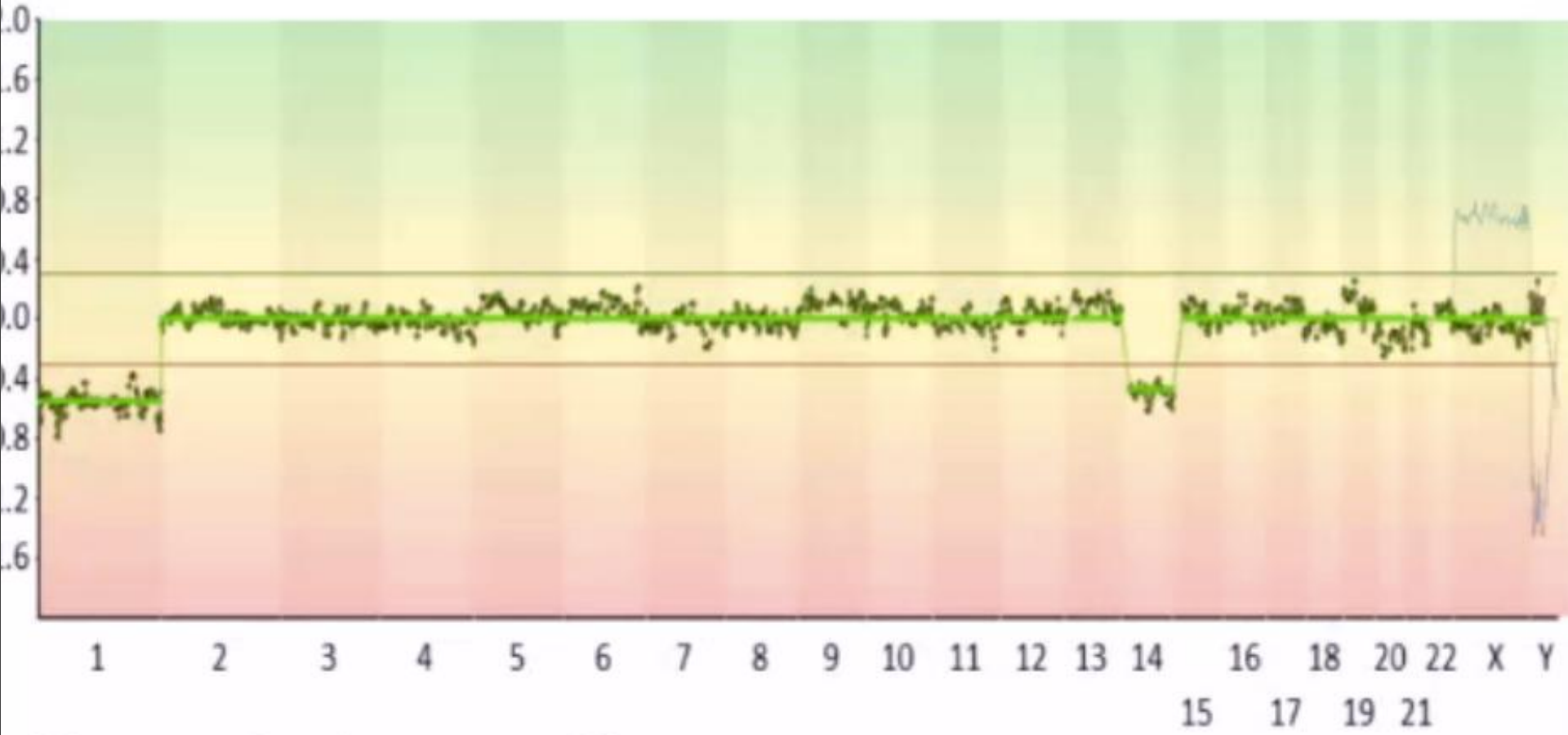
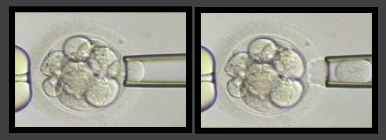
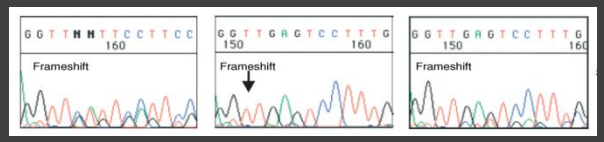
Microarray comparative genomic hybridization



Monosomy 1 and monosomy 14

Chromosome number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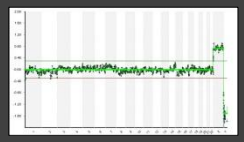
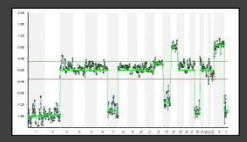
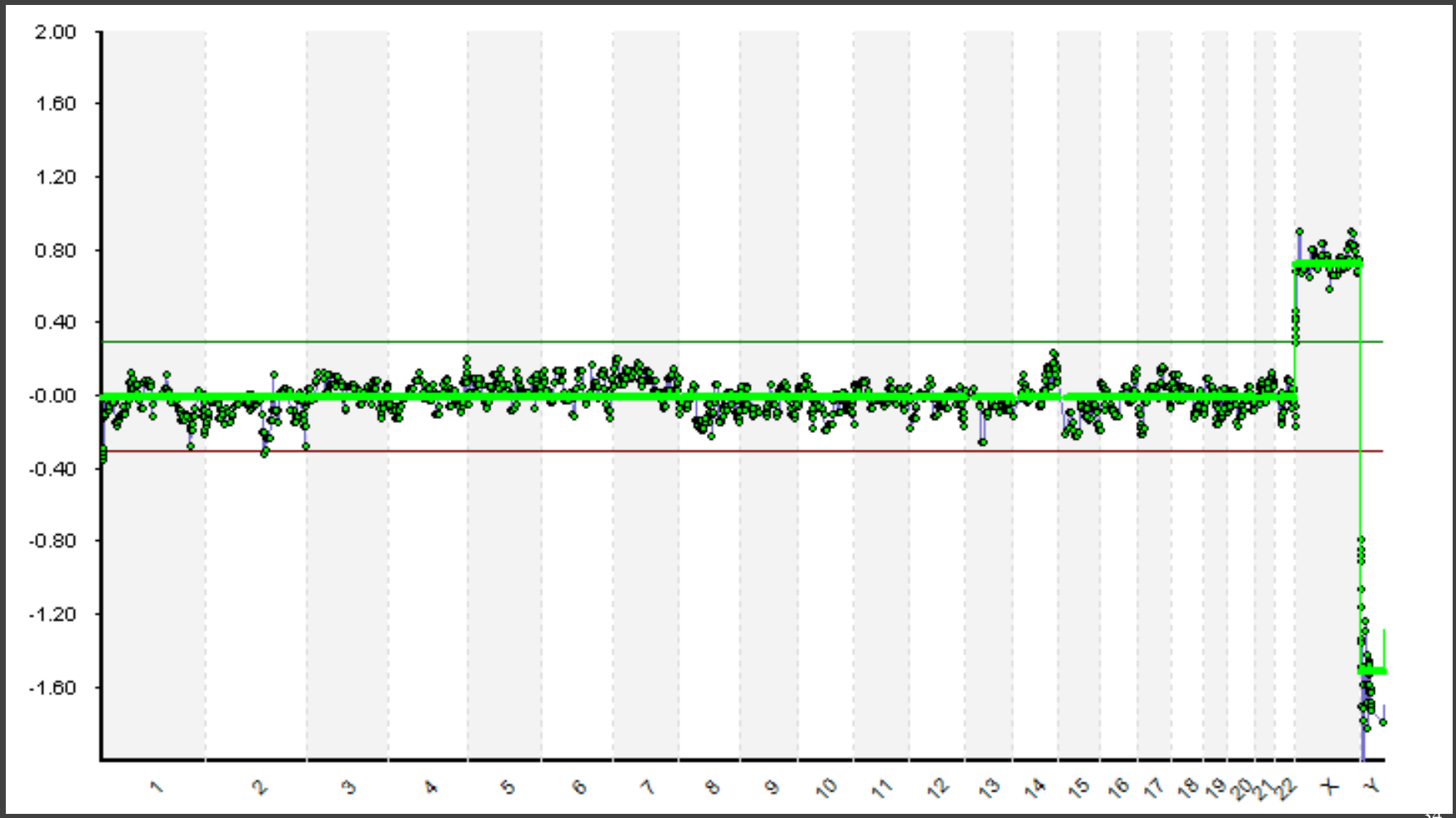



aCGH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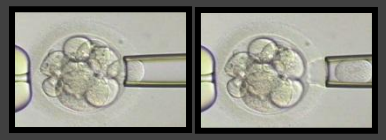

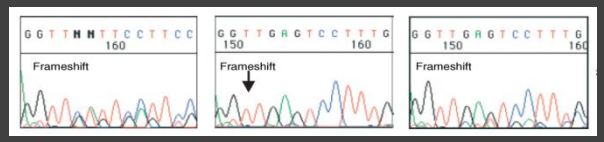
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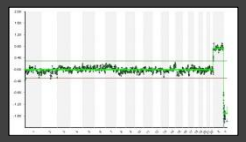

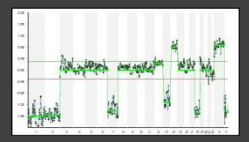

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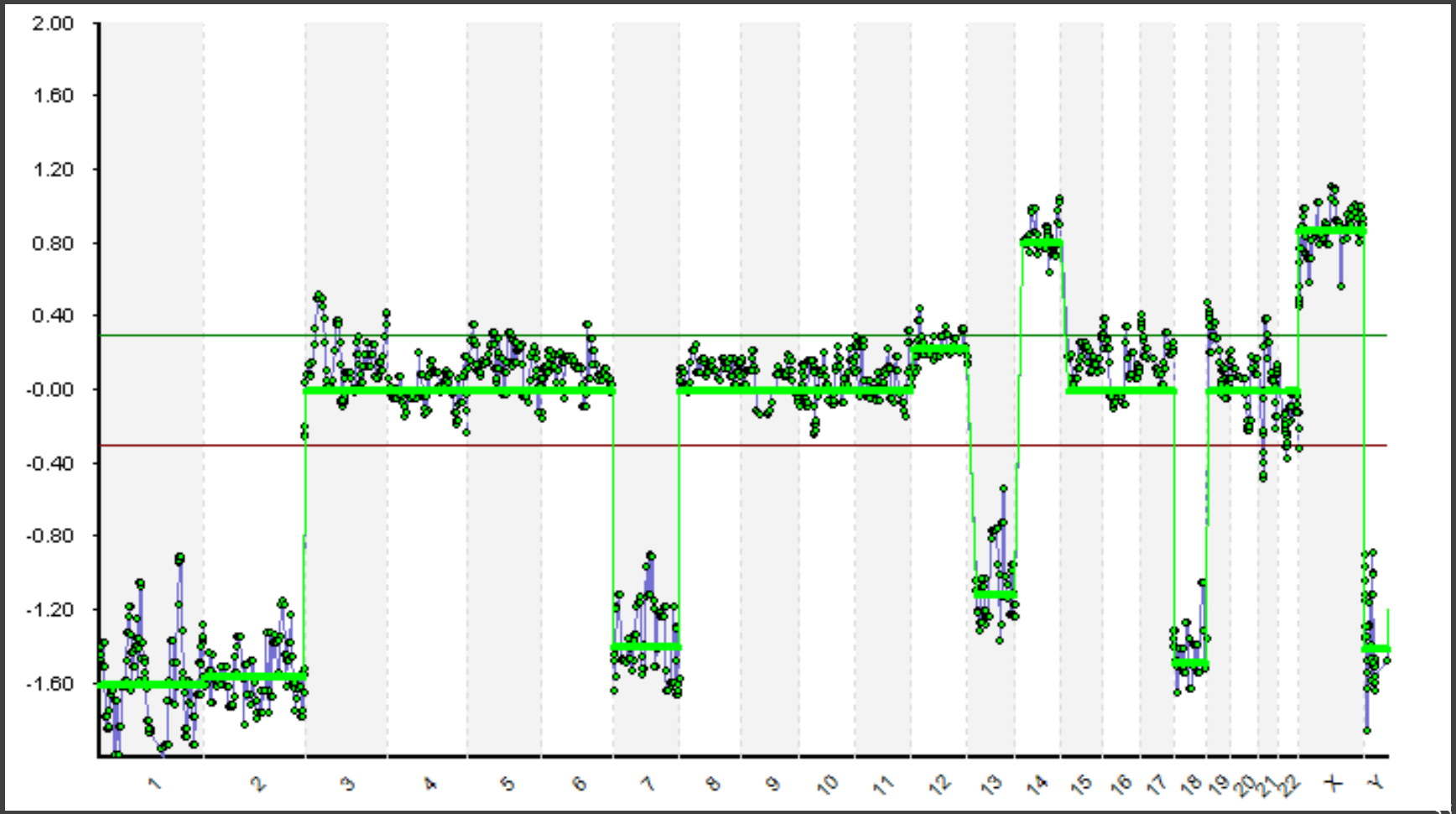
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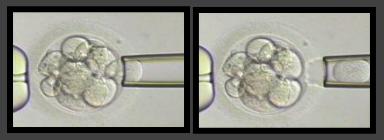
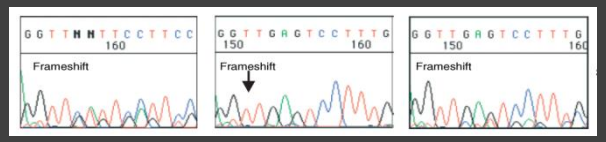
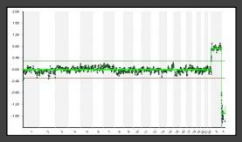
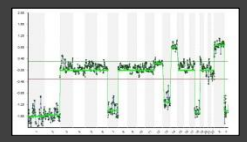
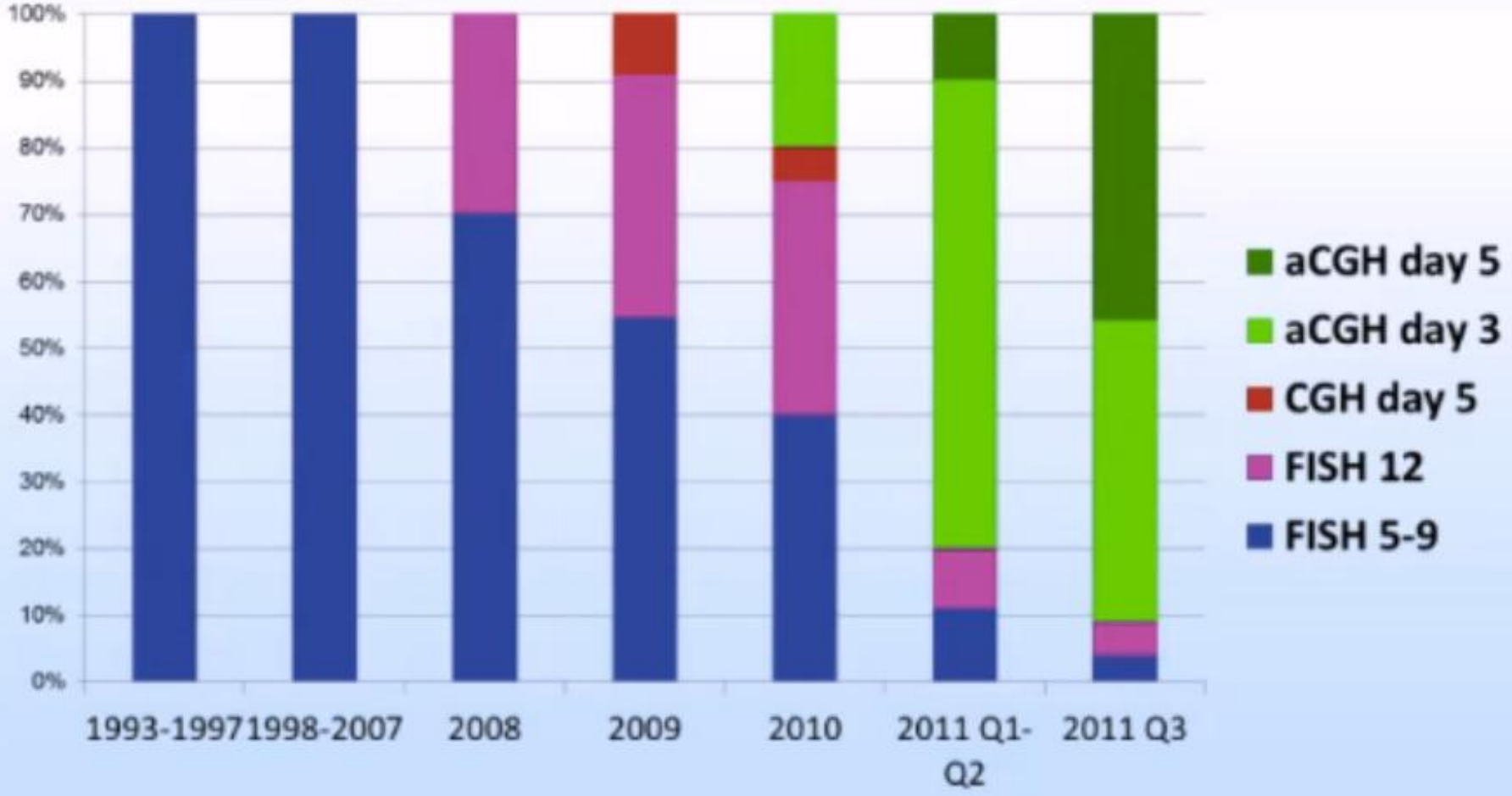



CGH

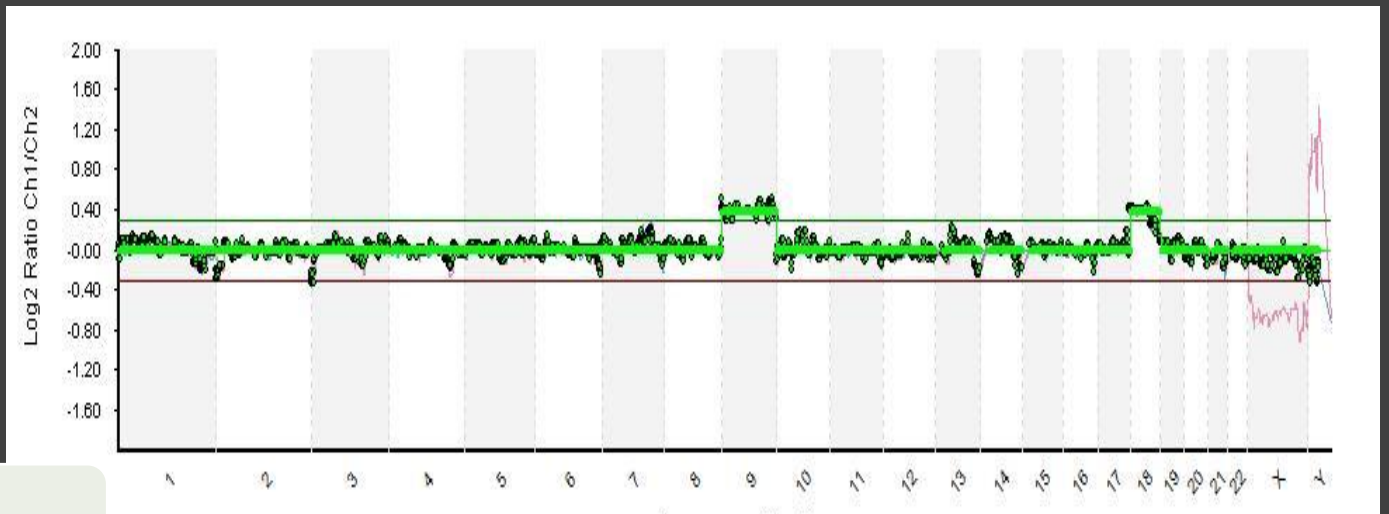
- Pros
  - Copy number information
  - All 24 chromosomes analysed
- Cons
  - Balanced translocation
  - Triploidy
  - Mosaicism
  - 72 hours hybridisation
  - Labour intensive
  - Expensive

PGD for aneuploidy: evolution



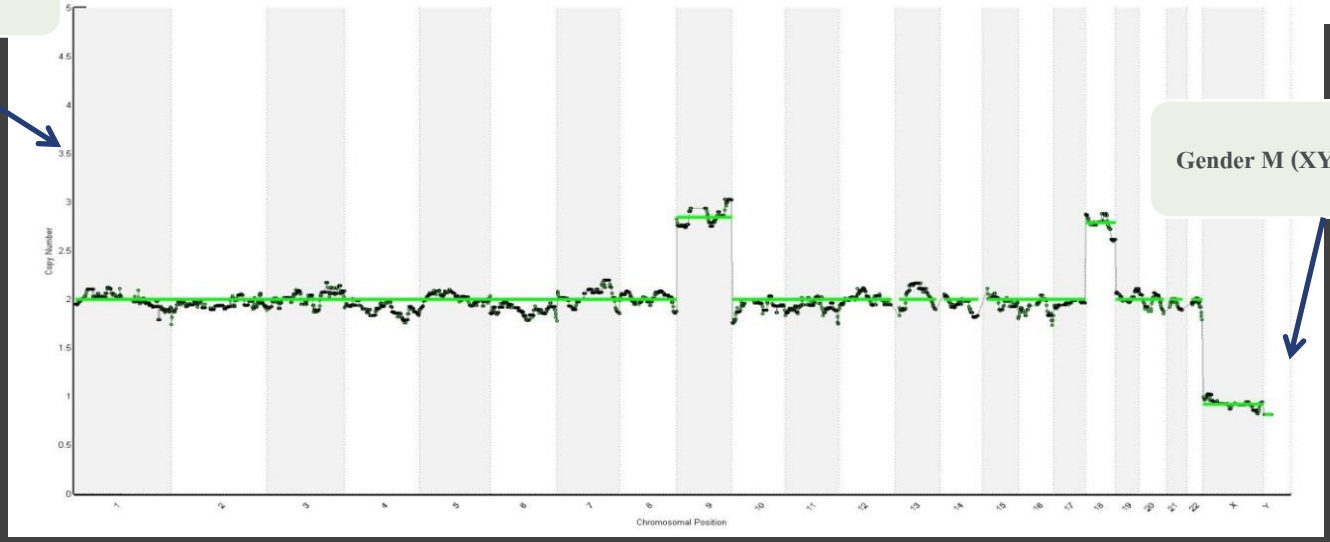
Performance Comparison between aCGH & NGS on Single Cell Day 3 Biopsy

aCGH



Copy number (not log2R)

NGS

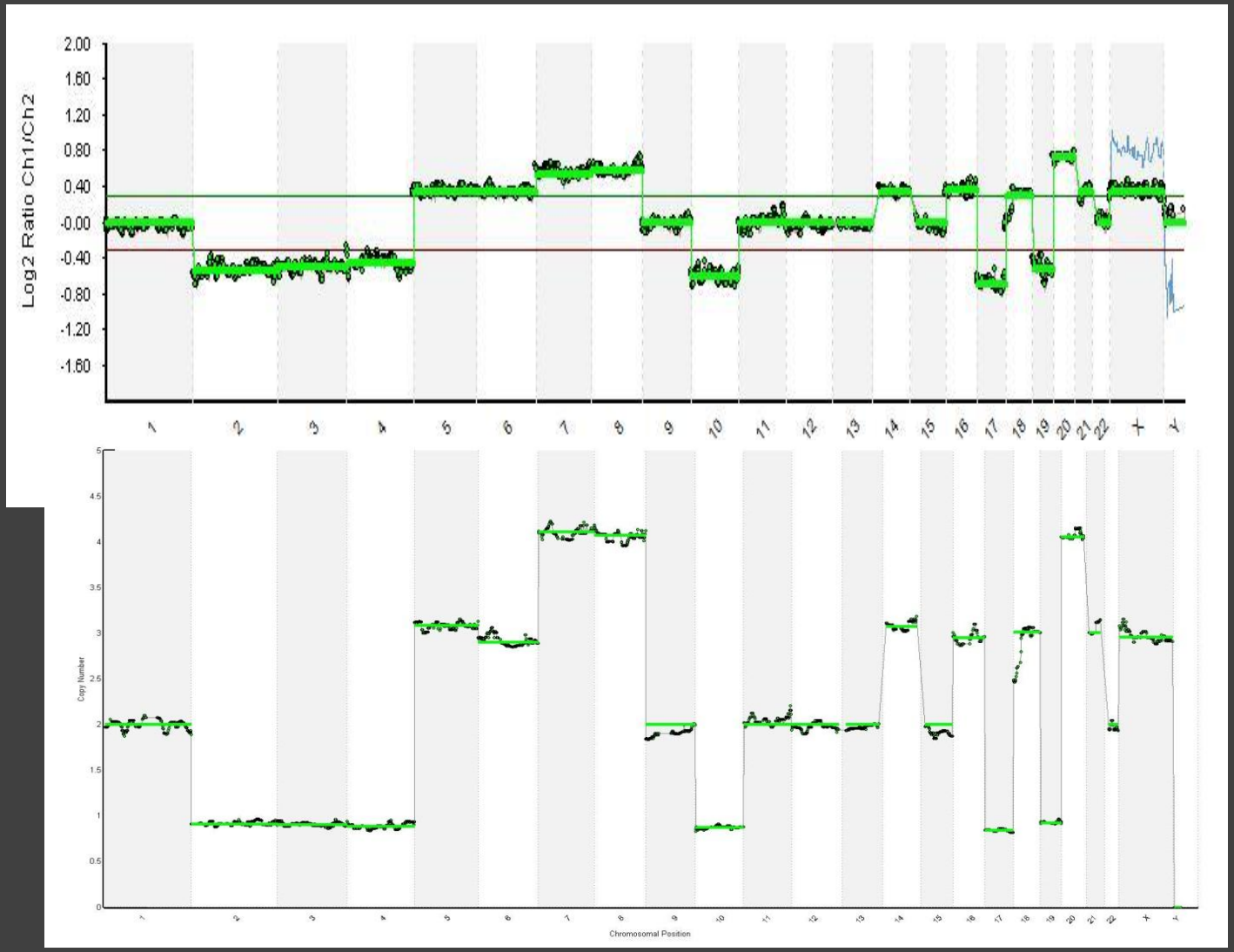


Gender M (XY) one copy of X & Y

Performance Comparison between aCGH & NGS on Complex Day 5 TE Biopsy

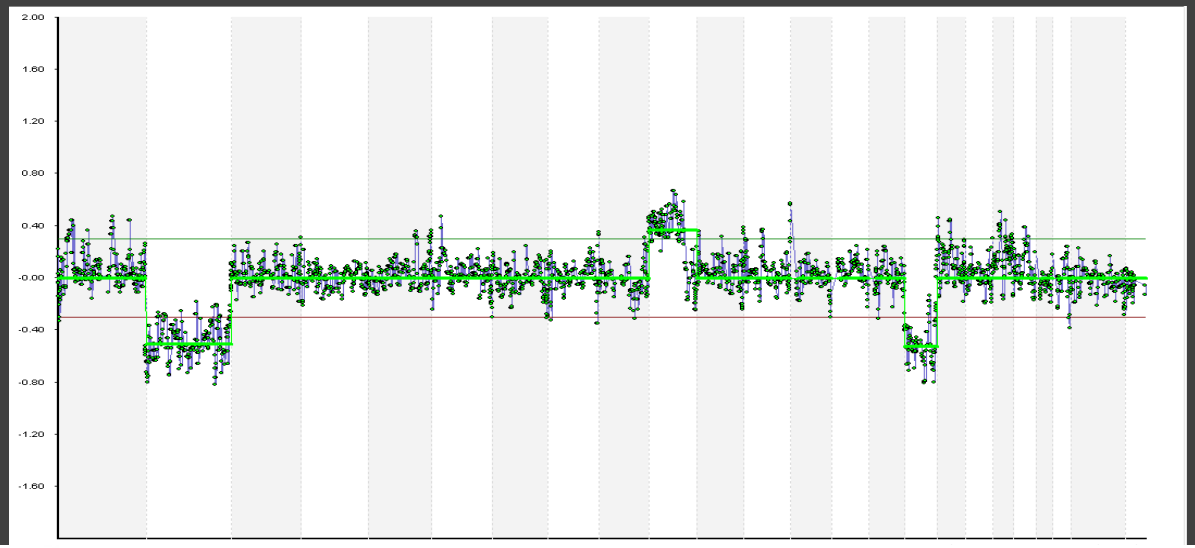
aCGH

NGS



Poorer Quality Amplification Products

aCGH



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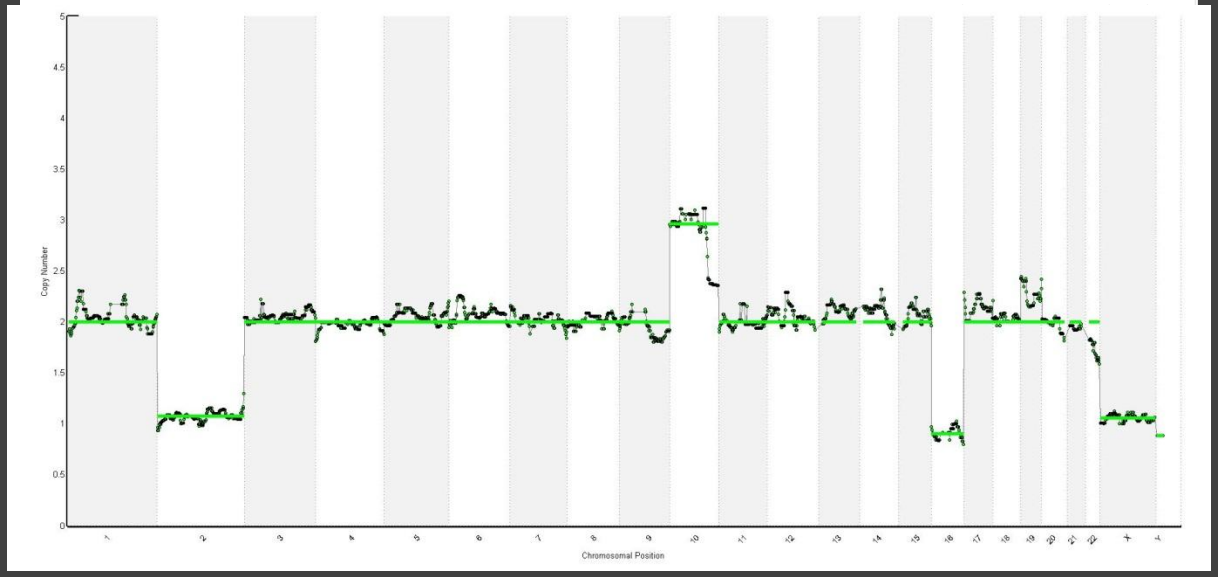
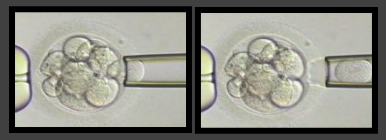
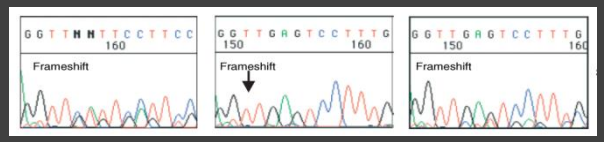
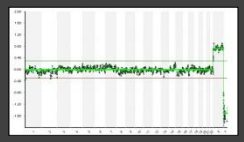
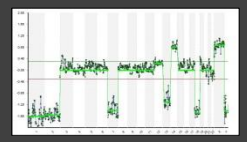
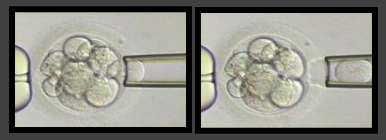
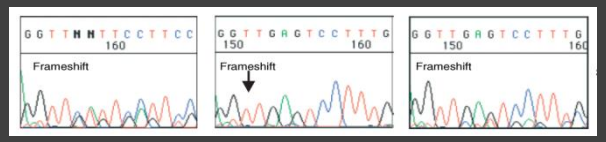


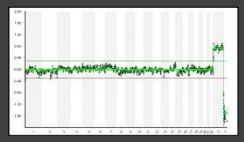
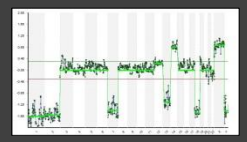



PREIMPLANTATION GENETIC DIAGNOSIS

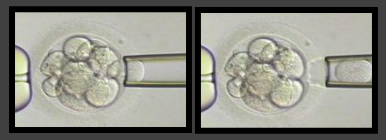
- Sample Retrieval Techniques
  - Polar body biopsy
  - Embryo biopsy*
  - Blastocyst biopsy
- Molecular Genetic Analyses
  - PCR* : Single Gene Disorders
  - FISH : Chromosome Abnormalities, Sexing
  - aCGH : Chromosome Abnormalities
  - Other techniques : sequencing, SNaPshot, SNP Array, NGS

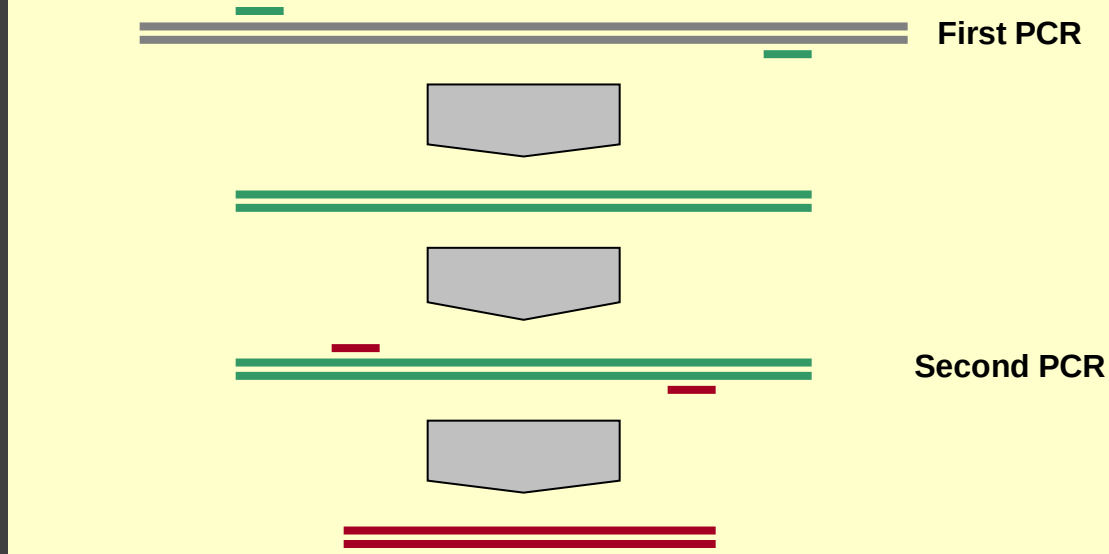



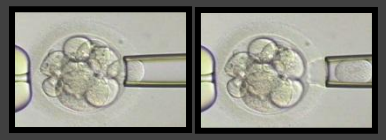
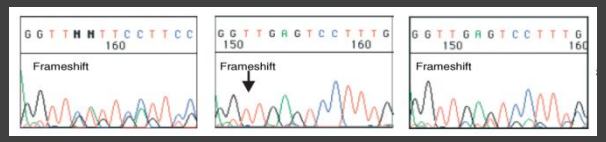
Single Cell PCR

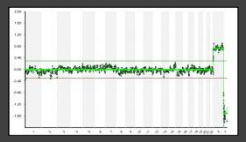
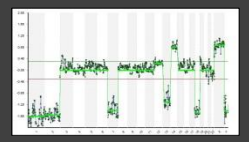
- 24 hours
- Amplification efficiency : nested PCR, fluorescent PCR
- Specific analysis technique for each mutation: deletion/insertion, substitution
- Contamination : PCR mixture preparation, STR markers
- Allele drop-out (ADO) : analyse 2 cells, linked markers



Nested PCR

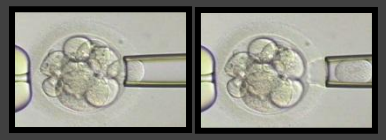


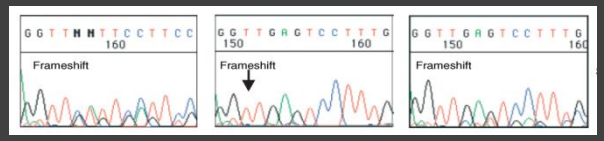



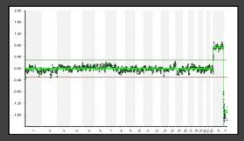



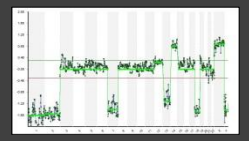
ANALYSIS TECHNIQUES

- Gel electrophoresis
- Restriction fragment length polymorphism (RFLP)
- Amplification refractory mutation system (ARMS)
- Heteroduplex analysis (HA)
- Single strand conformation polymorphism (SSCP)
- Denaturant gradient gel electrophoresis (DGGE)
- Fluorescent PCR (F-PCR)
- Sequencing
- Mini-sequencing (SNaPshot)
- Microarray (DNA chip), Karyomapping









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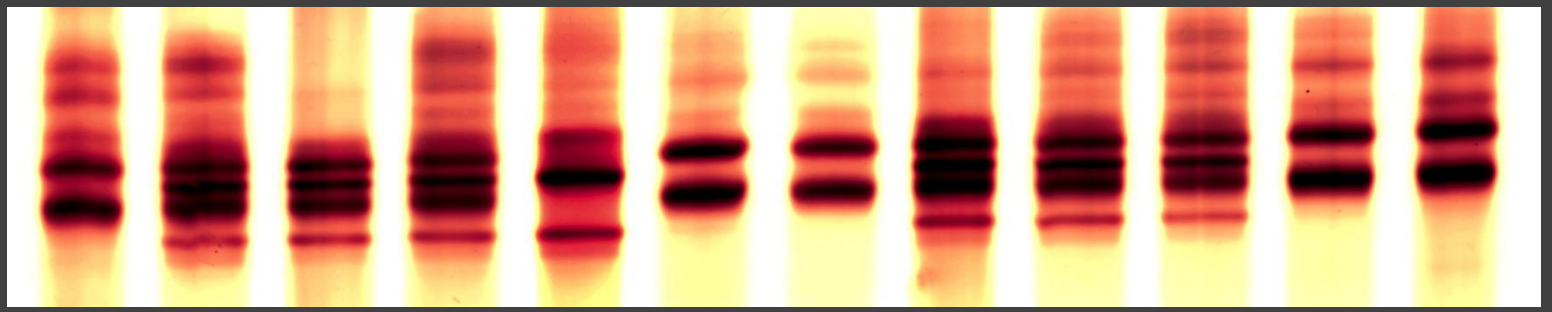

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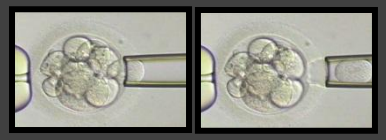

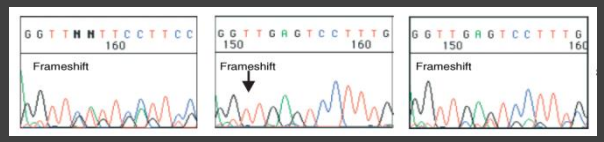
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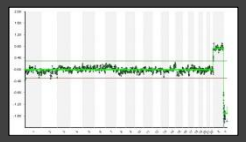

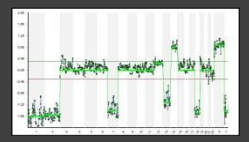

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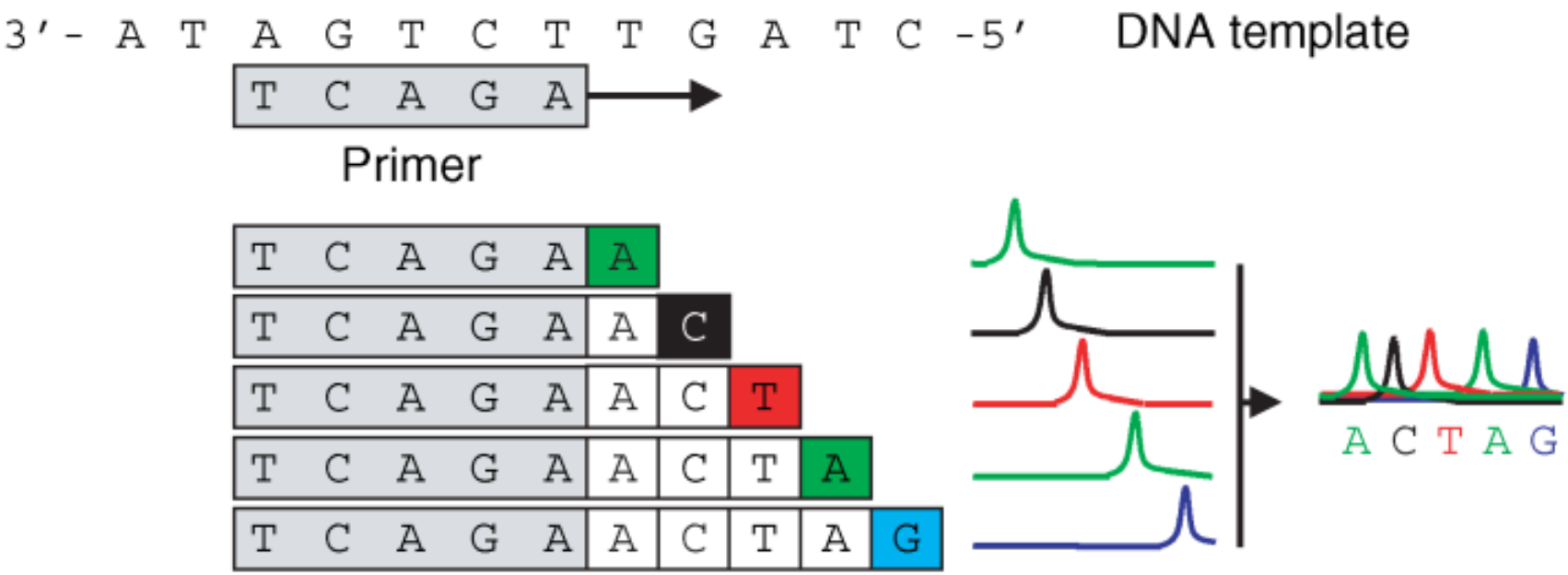
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Primer
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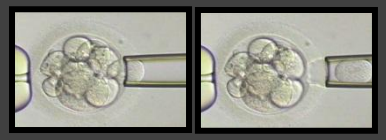
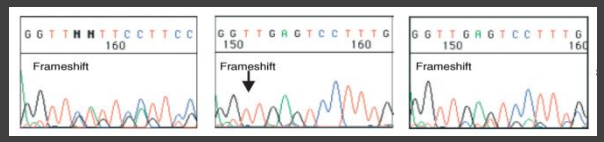
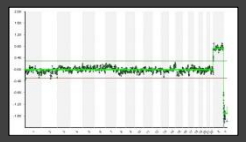
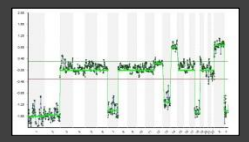
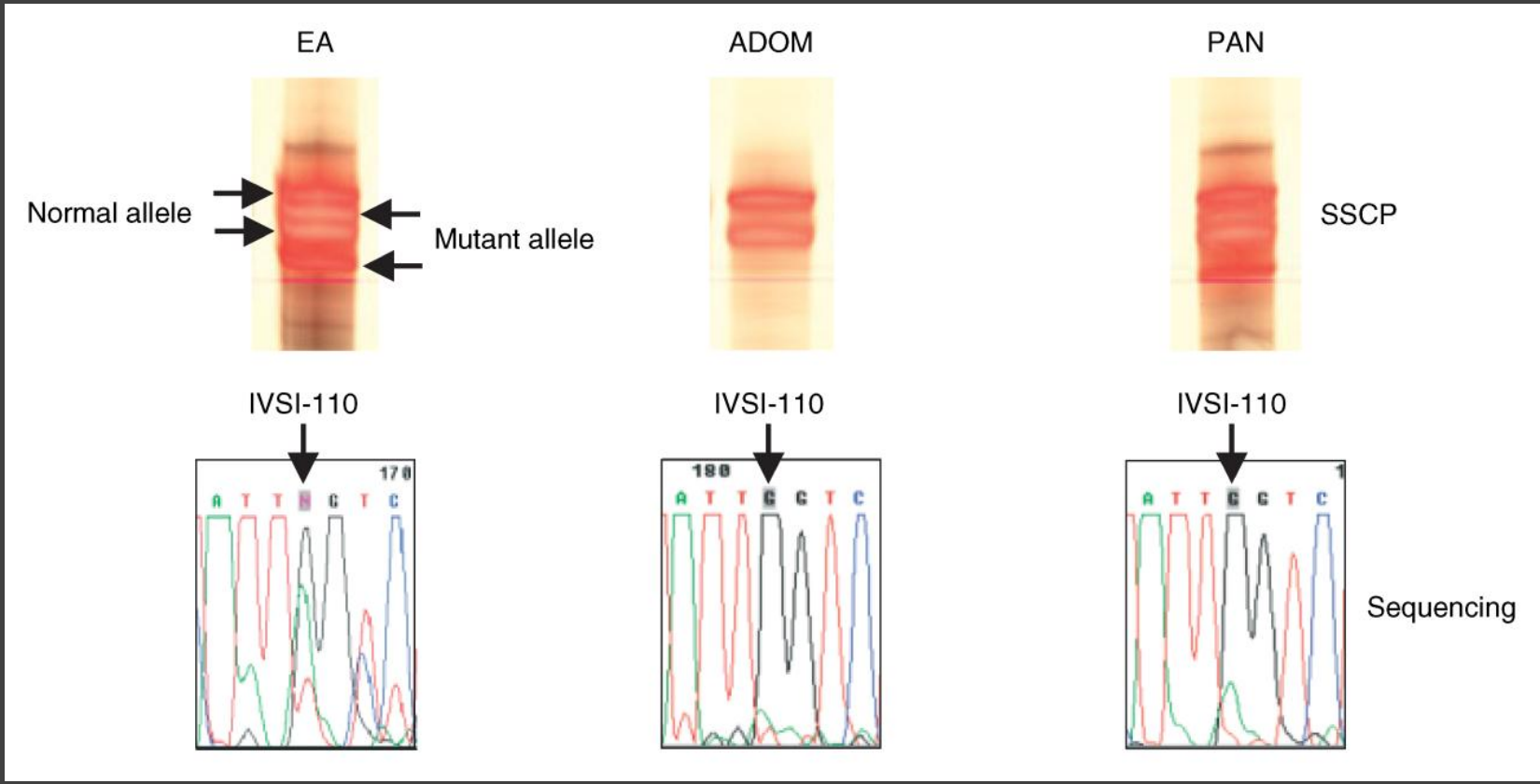


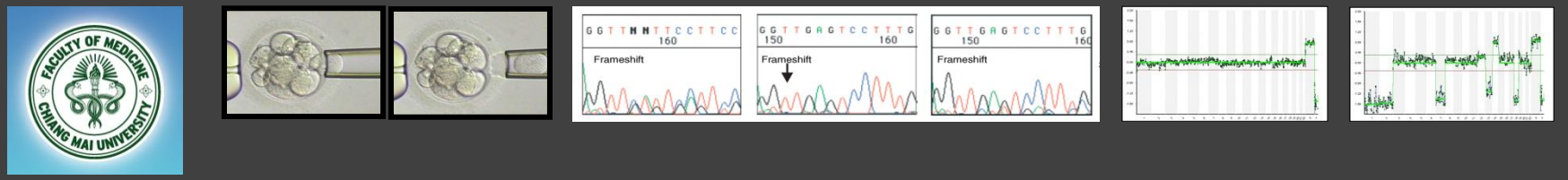






SINGLE CELL SEQUENCING
beta-Thalassemia IVSI-110 G>A





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