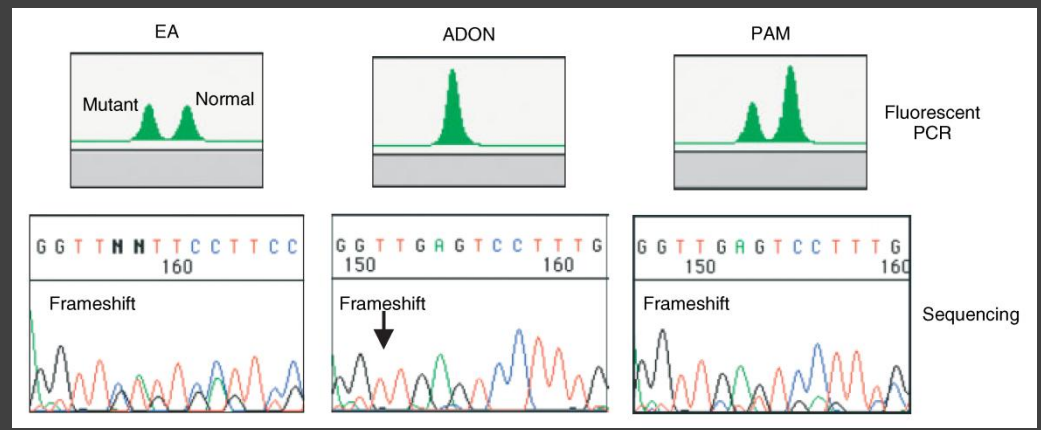
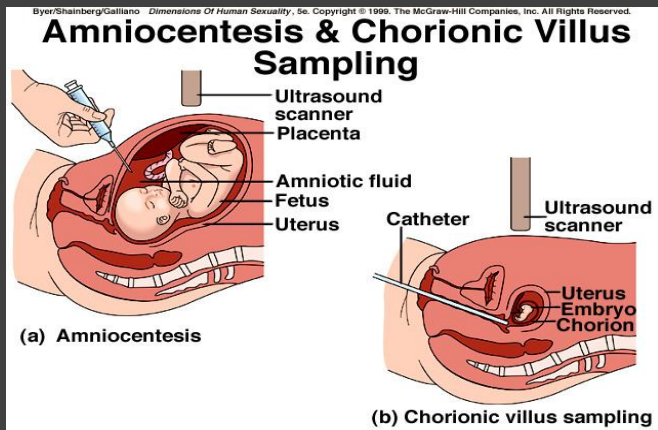
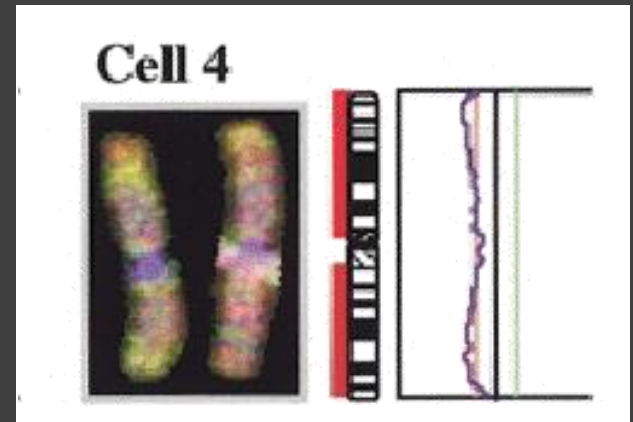
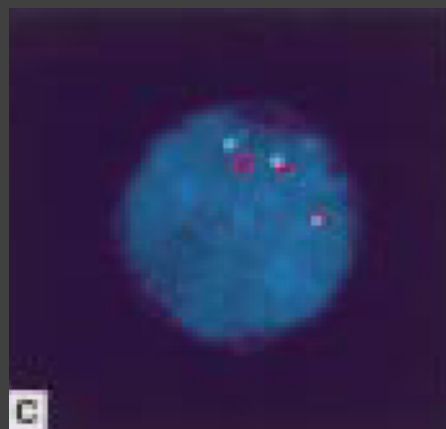
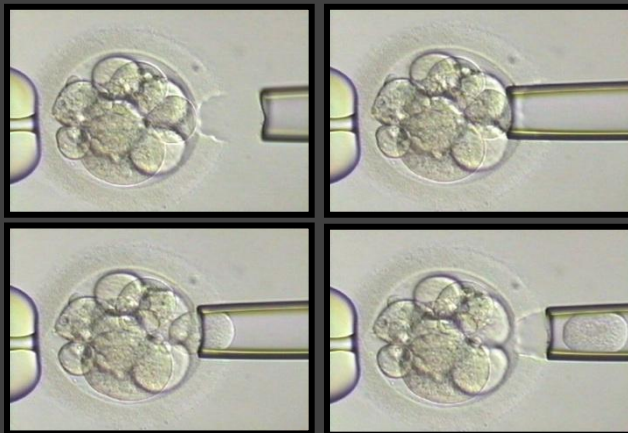
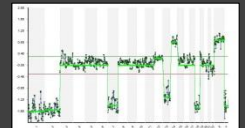
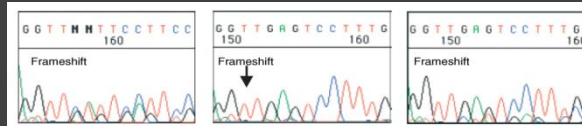




PREIMPLANTATION GENETIC DIAGNOSIS

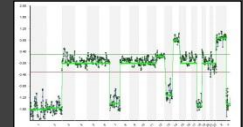
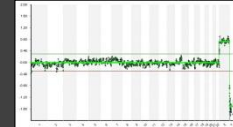
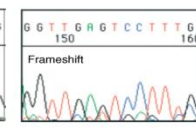
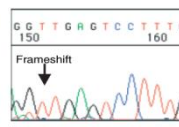
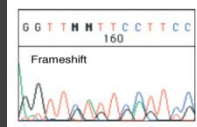
Wirawit Piyamongkol, MD, PhD
PGD Center, Department of OB&GYN,
Faculty of Medicine, Chiang Mai University





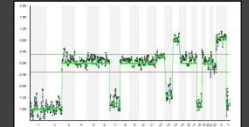
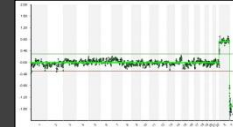
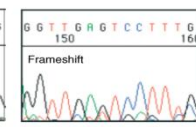
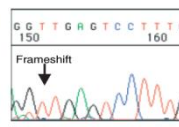
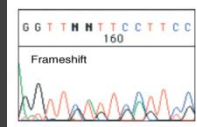
PREIMPLANTATION GENETIC DIAGNOSIS

- ❑ Designer baby, embryo selection
- ❑ Start a pregnancy with a disease free baby
- ❑ Eliminate the need for TOP
- ❑ Patients with moral or religious objections to TOP
- ❑ Subfertile couples carrying a chromosomal translocation or gonadal mosaicism
- ❑ Carriers of inherited disease can avoid repeated TOP
- ❑ Patients at risk of having offspring with late onset or non-life-threatening diseases

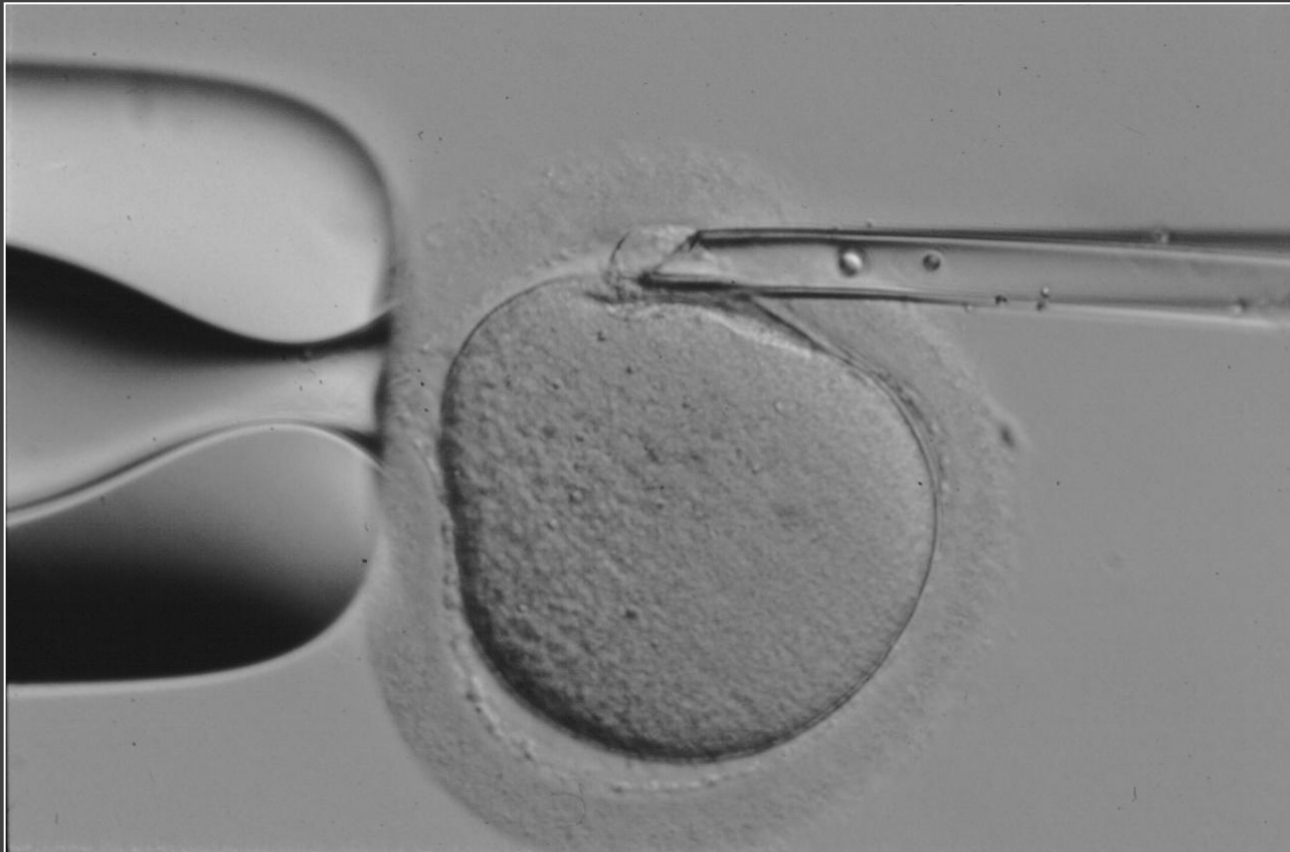


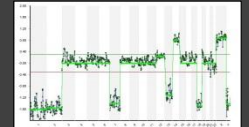
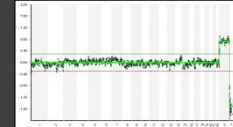
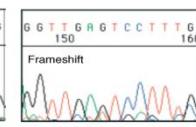
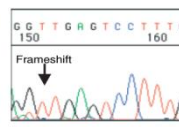
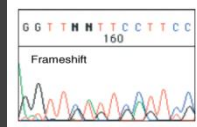
PREIMPLANTATION GENETIC DIAGNOSIS

- **Sample Retrieval Techniques**
 - Polar body biopsy
 - Embryo biopsy*
 - Blastocyst biopsy



POLAR BODY BIOPSY





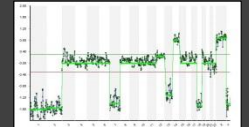
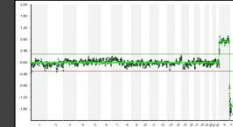
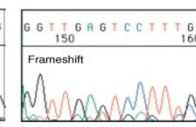
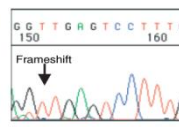
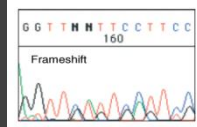
POLAR BODY BIOPSY

■ Pros

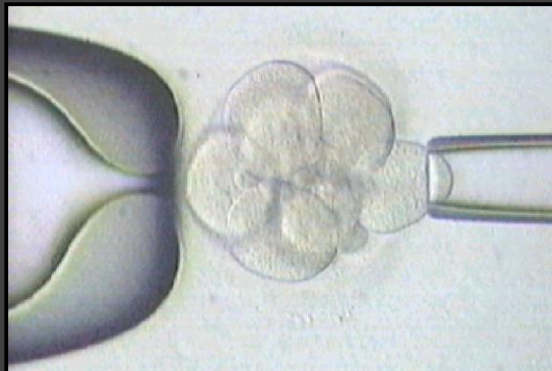
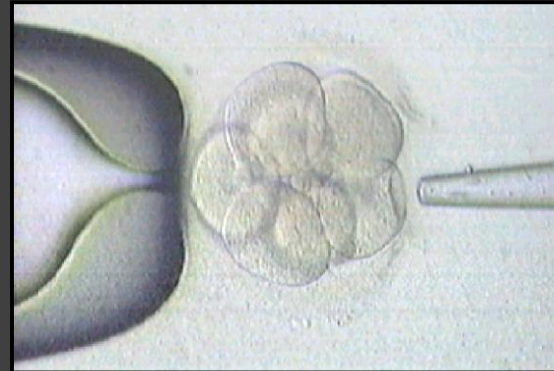
- Preconception diagnosis
- Harmless
- Un-used part

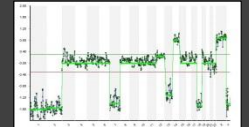
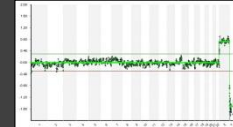
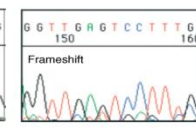
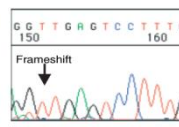
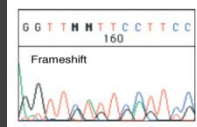
■ Cons

- Labor intensive
- Chance of recombination
- Mutant embryos in recessive disorders
- Paternal allele
- Post-zygotic event

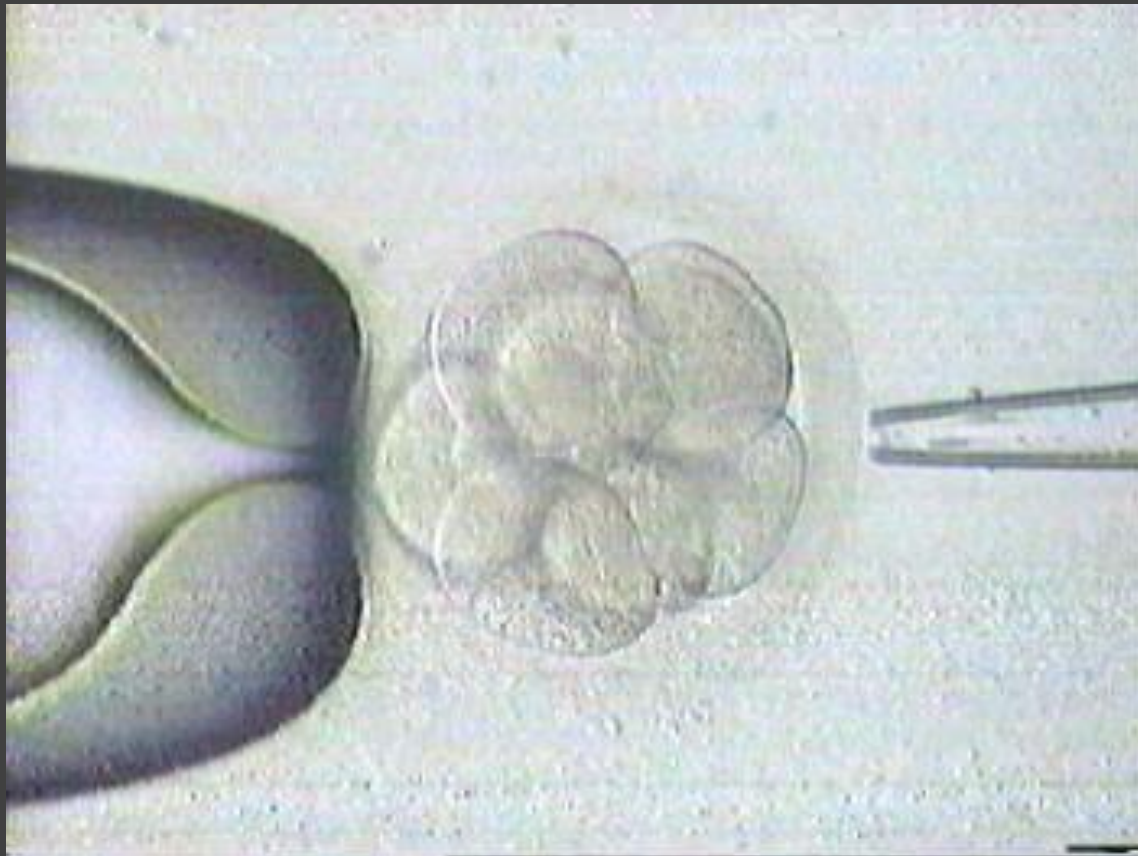


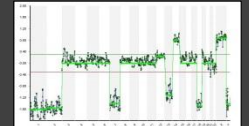
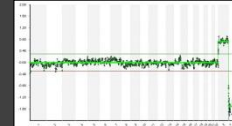
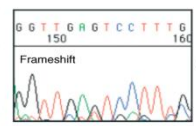
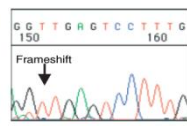
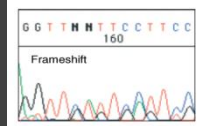
CLEAVAGE STAGE EMBRYO BIOPSY





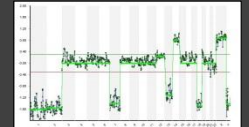
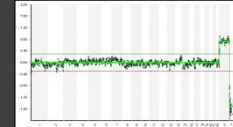
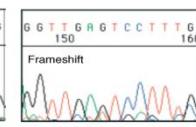
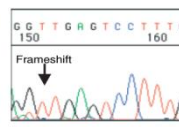
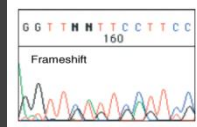
CLEAVAGE STAGE EMBRYO BIOPSY





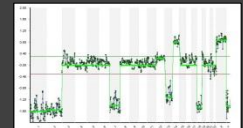
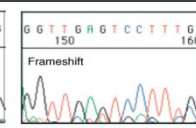
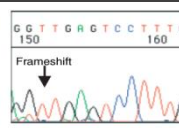
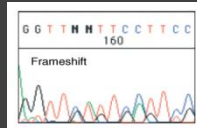
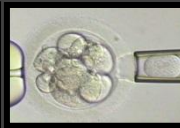
CLEAVAGE STAGE EMBRYO BIOPSY





CLEAVAGE STAGE EMBRYO BIOPSY





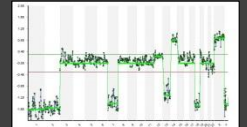
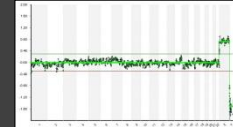
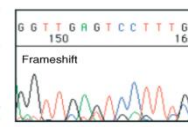
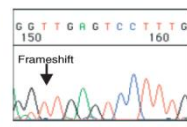
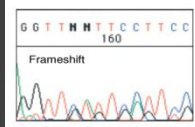
CLEAVAGE STAGE EMBRYO BIOPSY

■ Pros

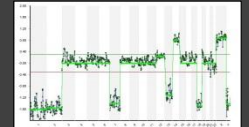
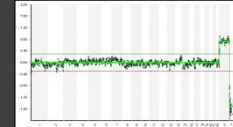
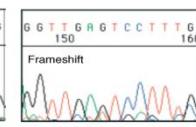
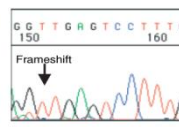
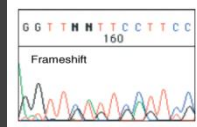
- Totipotency
- Embryonic development

■ Cons

- 1-2 cells for analysis
- Amplification efficiency
- Contamination
- Allele drop out (ADO)
- 24 hours



b



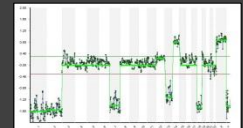
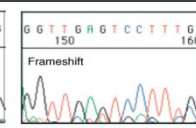
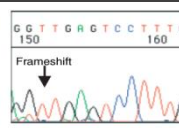
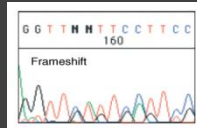
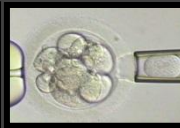
BLASTOCYST BIOPSY

■ Pros

- 10-30 cells for analysis
- No. of embryos for diagnosis

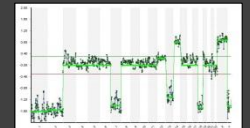
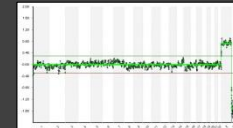
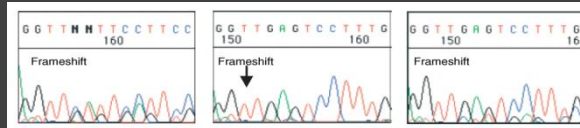
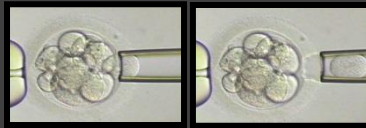
■ Cons

- No. of surviving embryos
- Chance of mosaicism
- Endometrium

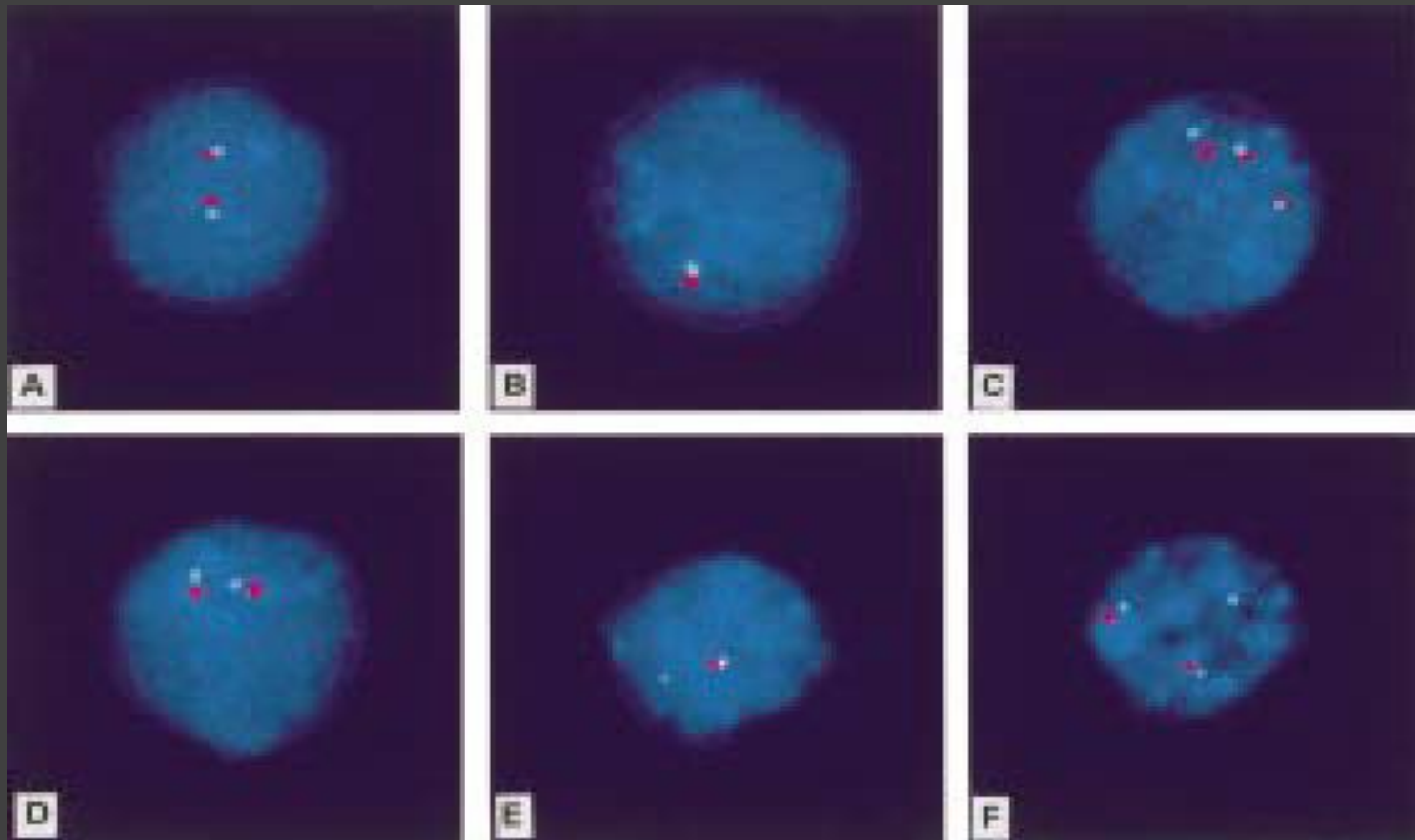


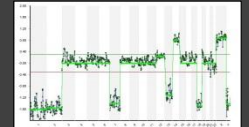
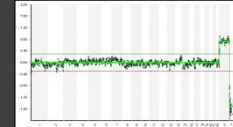
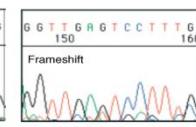
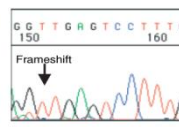
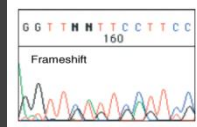
PREIMPLANTATION GENETIC DIAGNOSIS

- **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



FLUORESCENT IN SITU HYBRIDISATION (FISH)





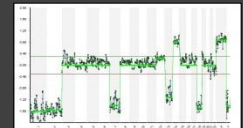
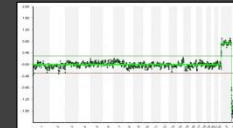
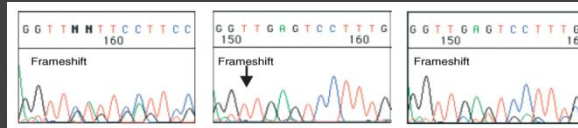
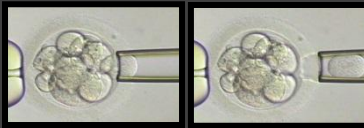
FISH

■ Pros

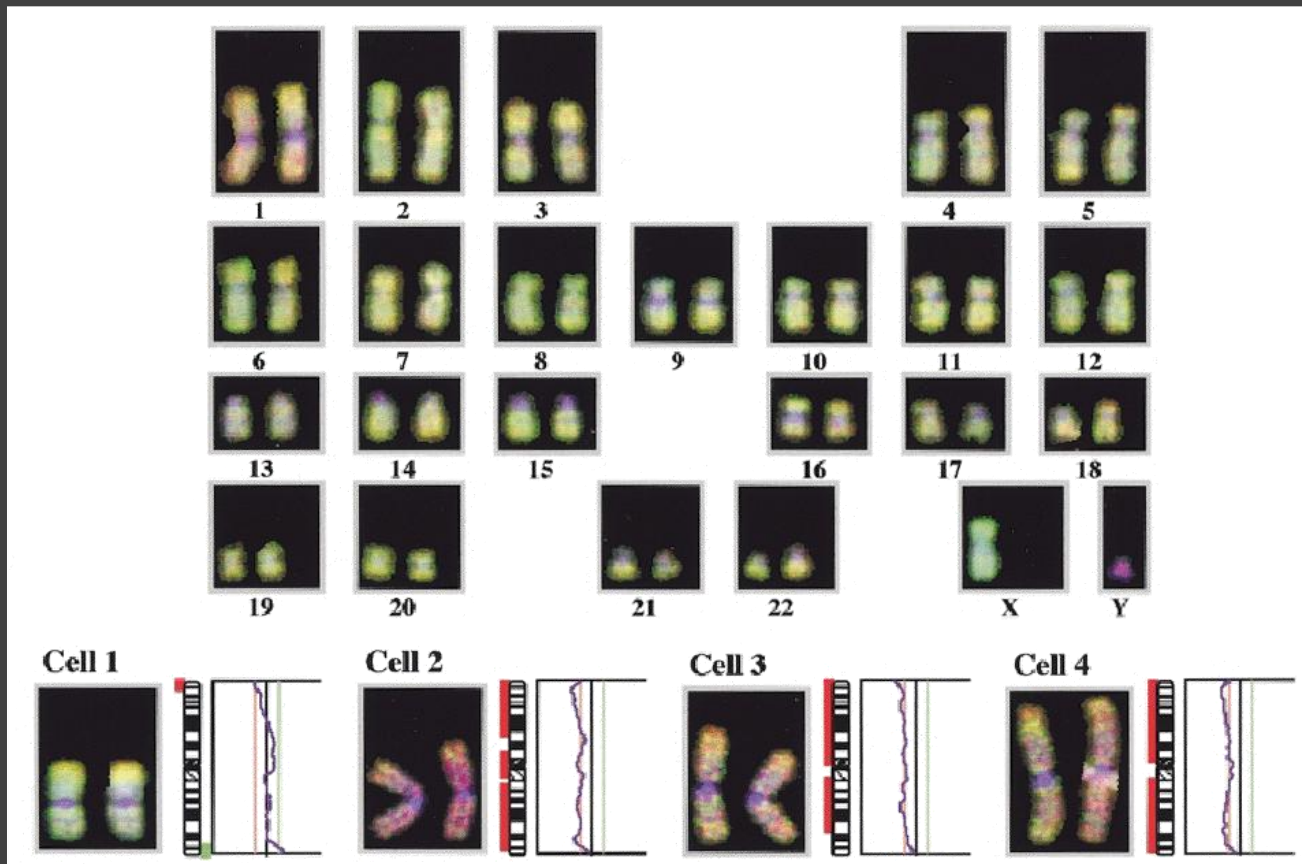
- Copy number information
- No contamination problem

■ Cons

- Limited No. of chromosomes analysed
- Hybridisation efficiency
- Overlapping signals
- Split signals

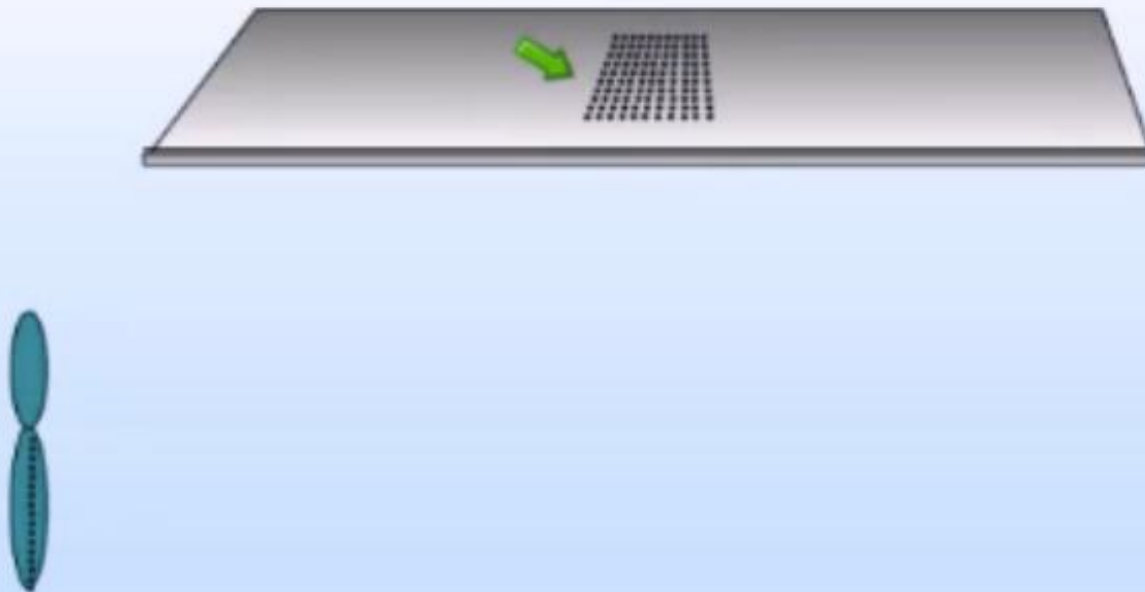


COMPARATIVE GENOME HYBRIDISATION (CGH)



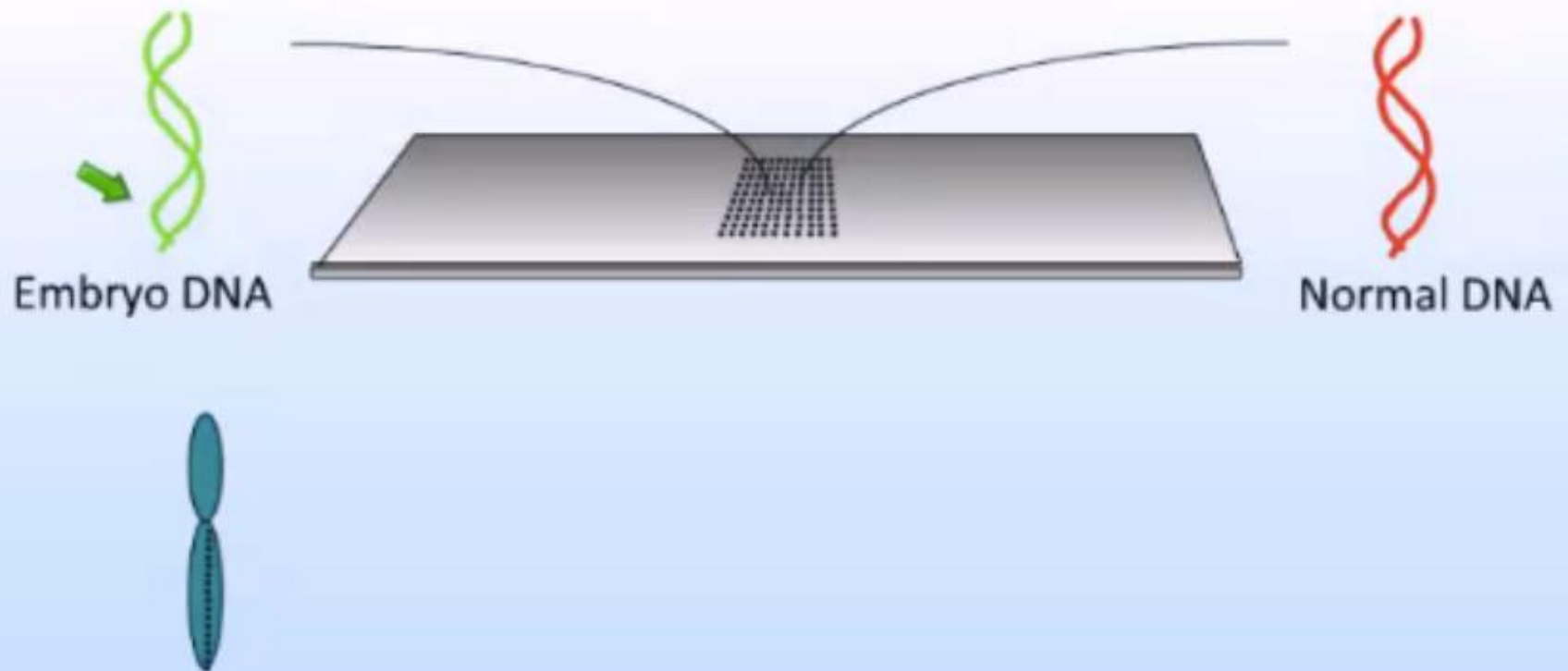
Microarray comparative genomic hybridization

- Rapid – results in 12-24 hours
- Allows the copy number of every chromosome to be determined



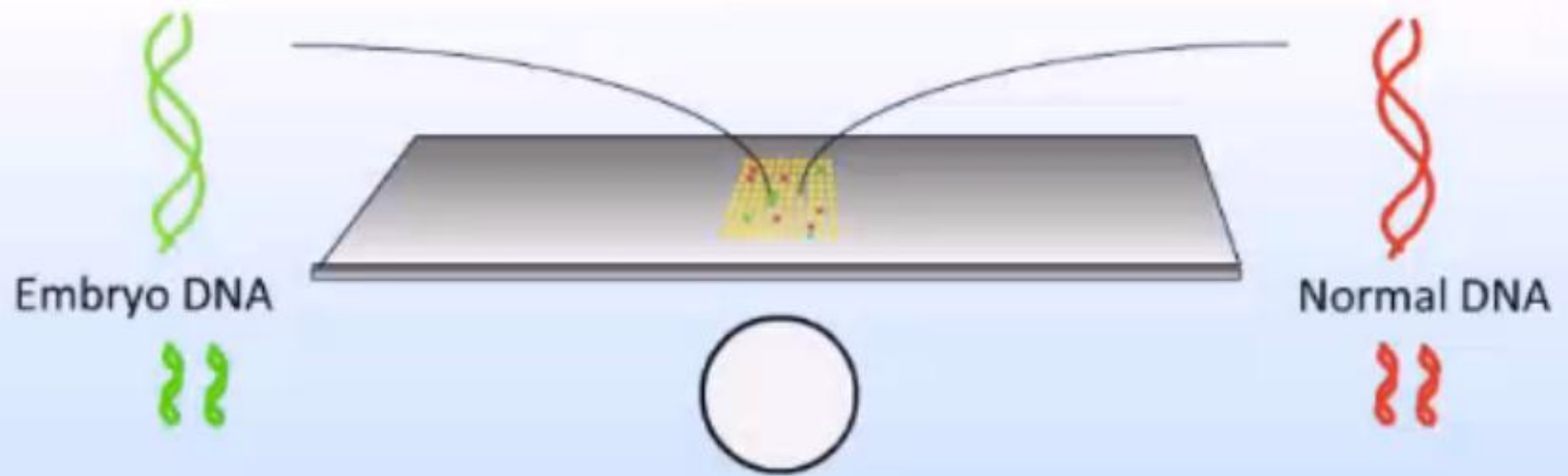
Microarray comparative genomic hybridization

- Rapid – results in 12-24 hours
- Allows the copy number of every chromosome to be determined



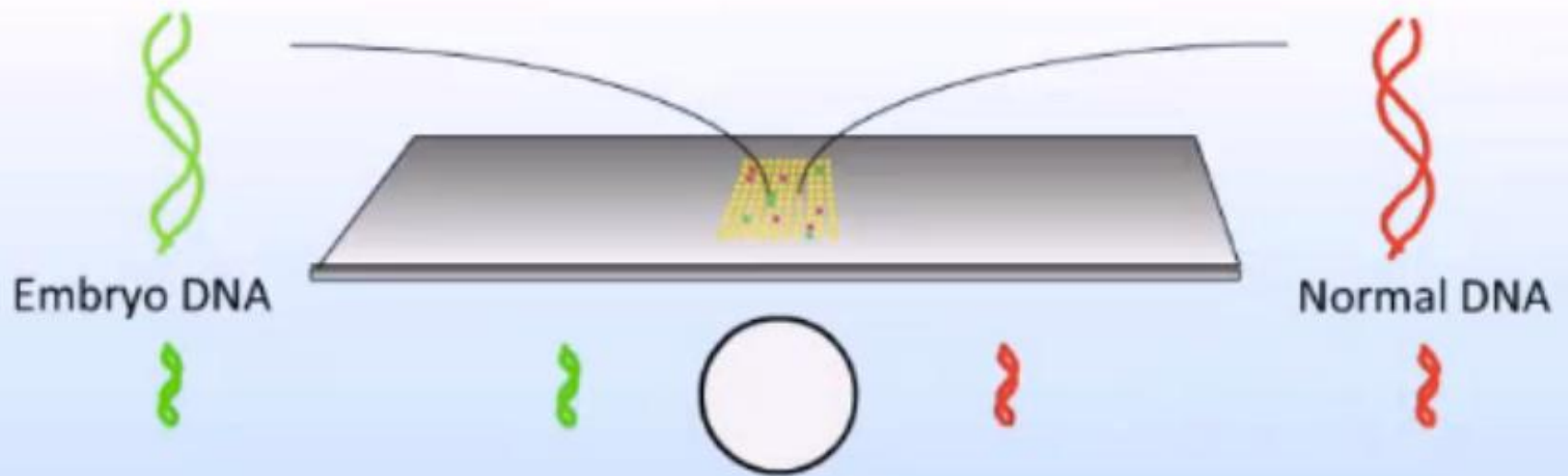
Microarray comparative genomic hybridization

- Rapid – results in 12-24 hours
- Allows the copy number of every chromosome to be determined



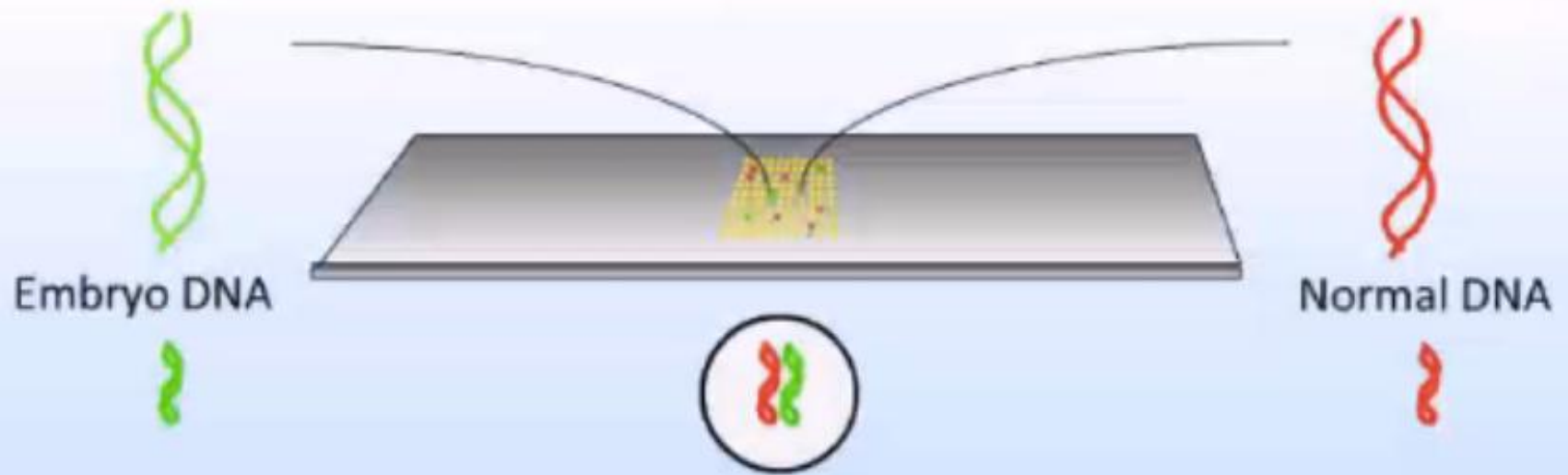
Microarray comparative genomic hybridization

- Rapid – results in 12-24 hours
- Allows the copy number of every chromosome to be determined



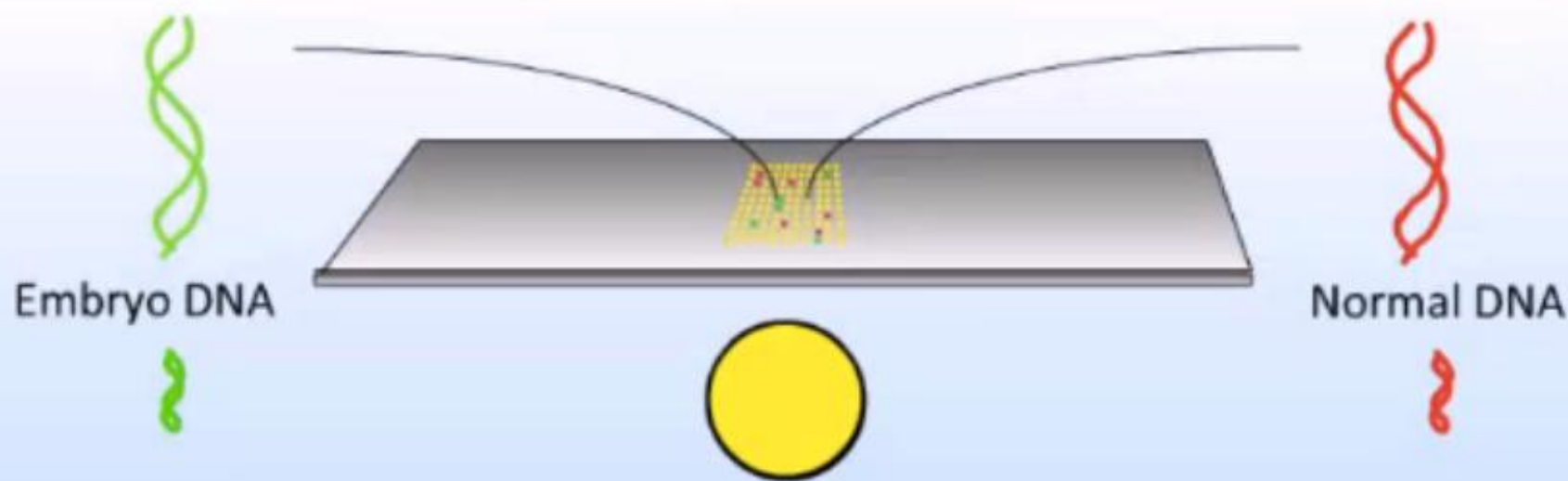
Microarray comparative genomic hybridization

- Rapid – results in 12-24 hours
- Allows the copy number of every chromosome to be determined



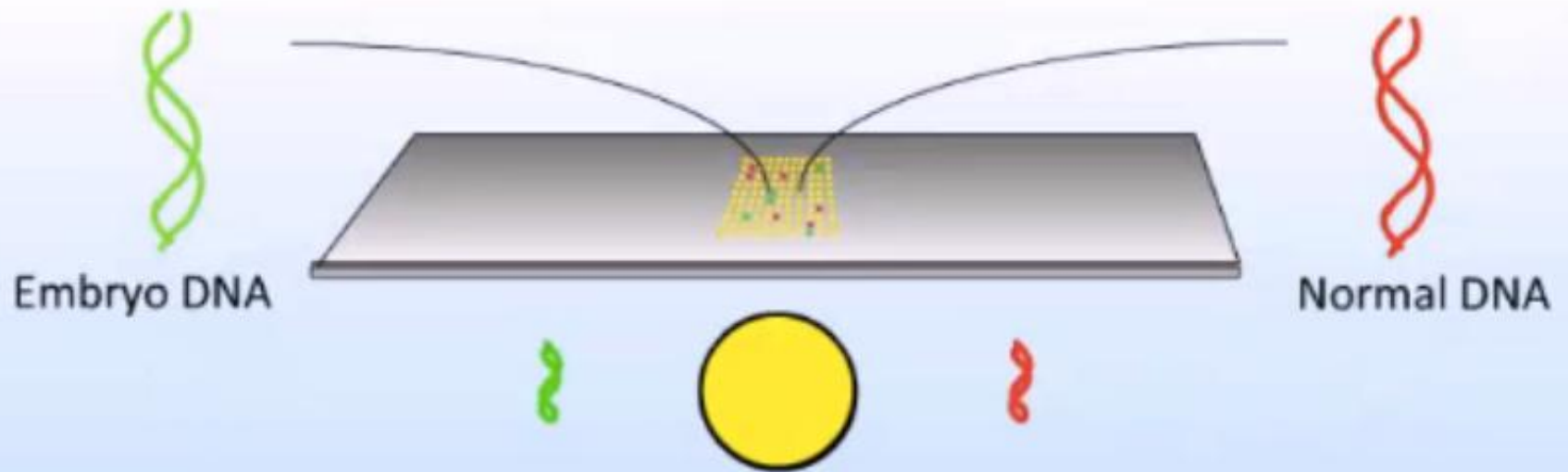
Microarray comparative genomic hybridization

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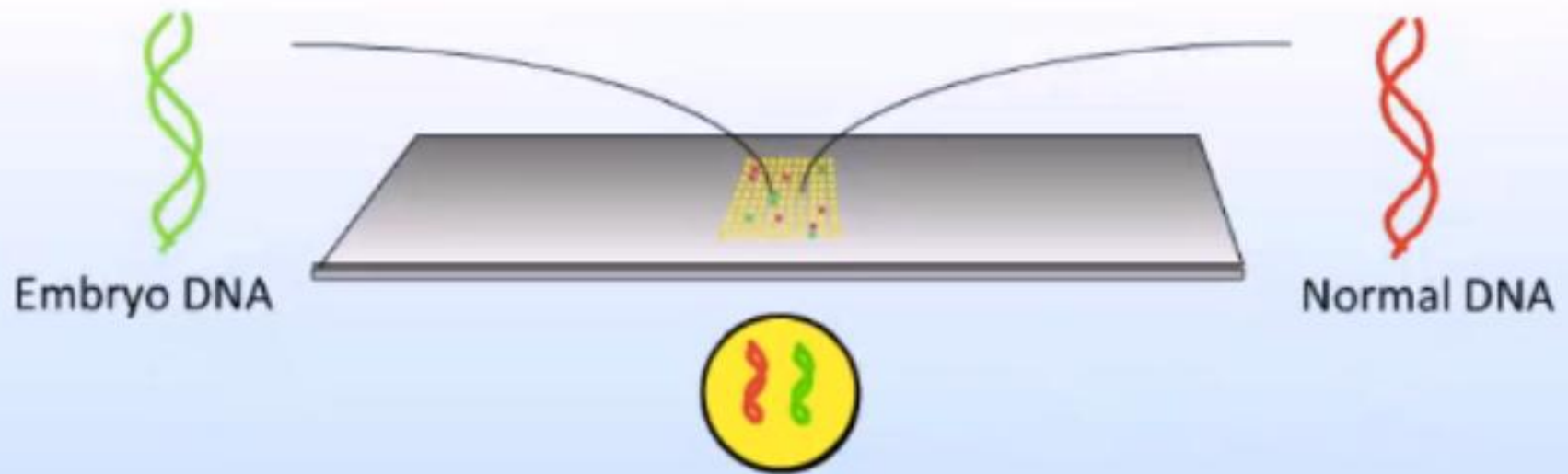
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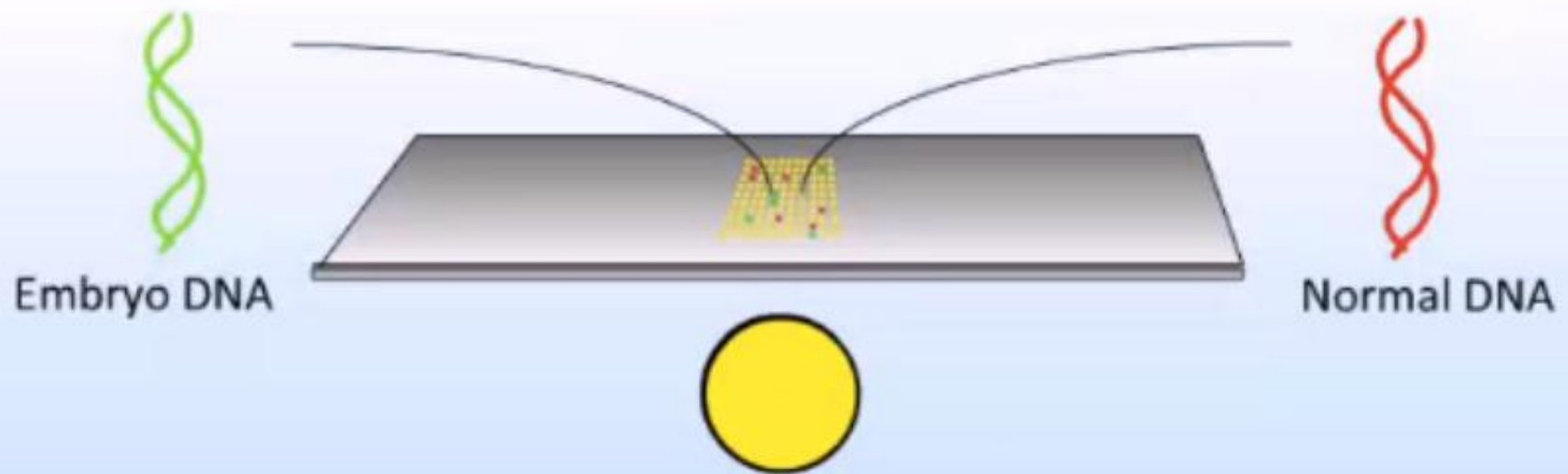
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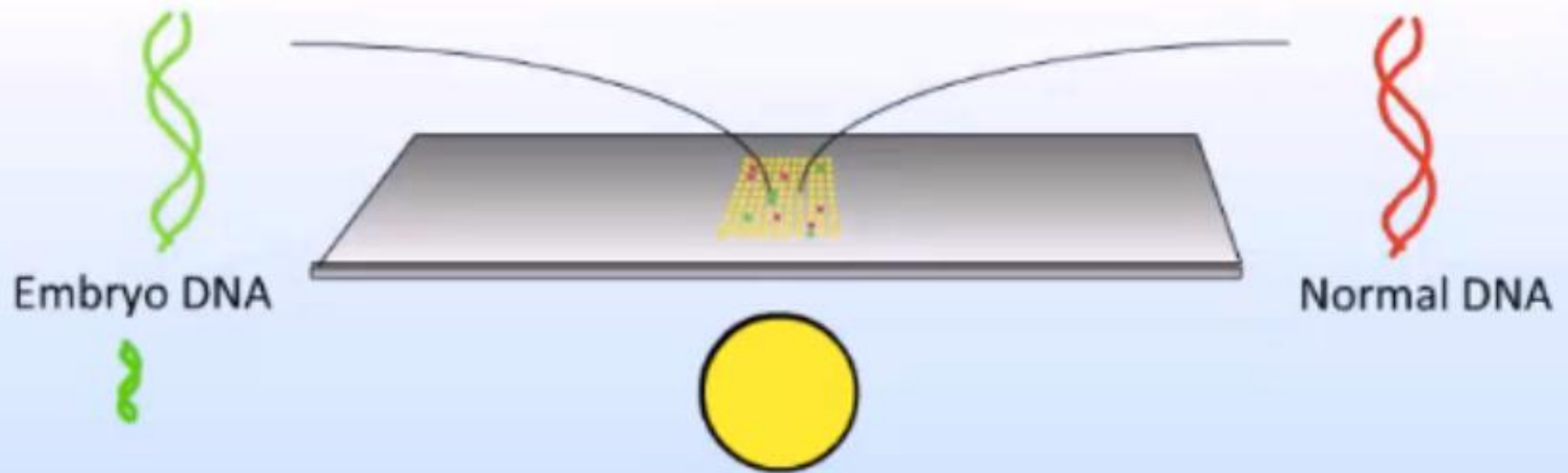
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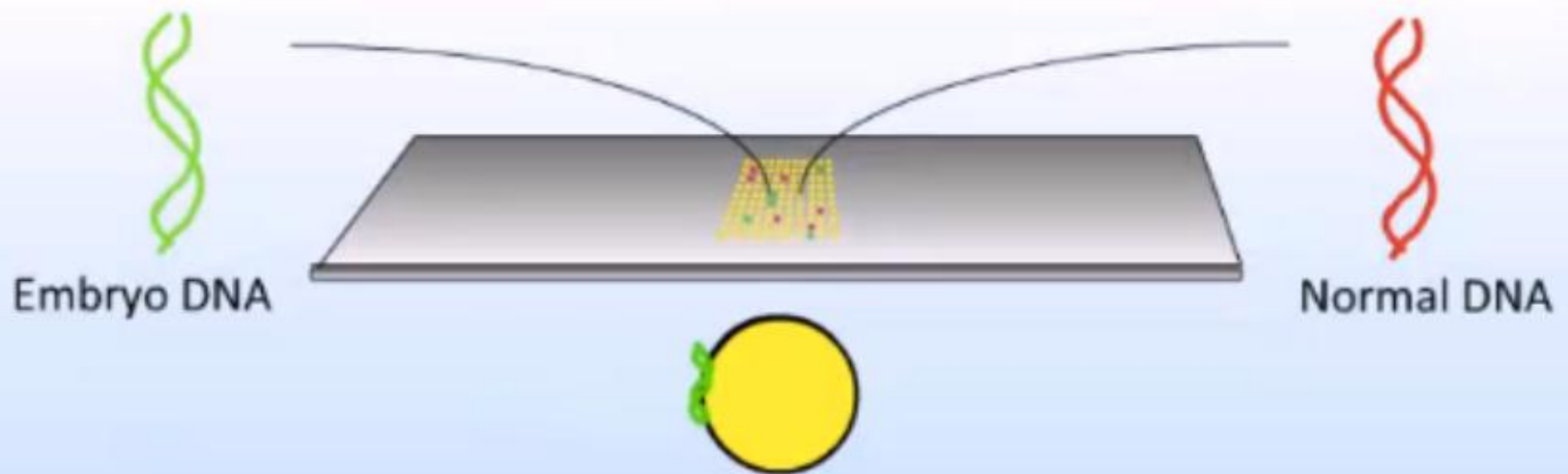
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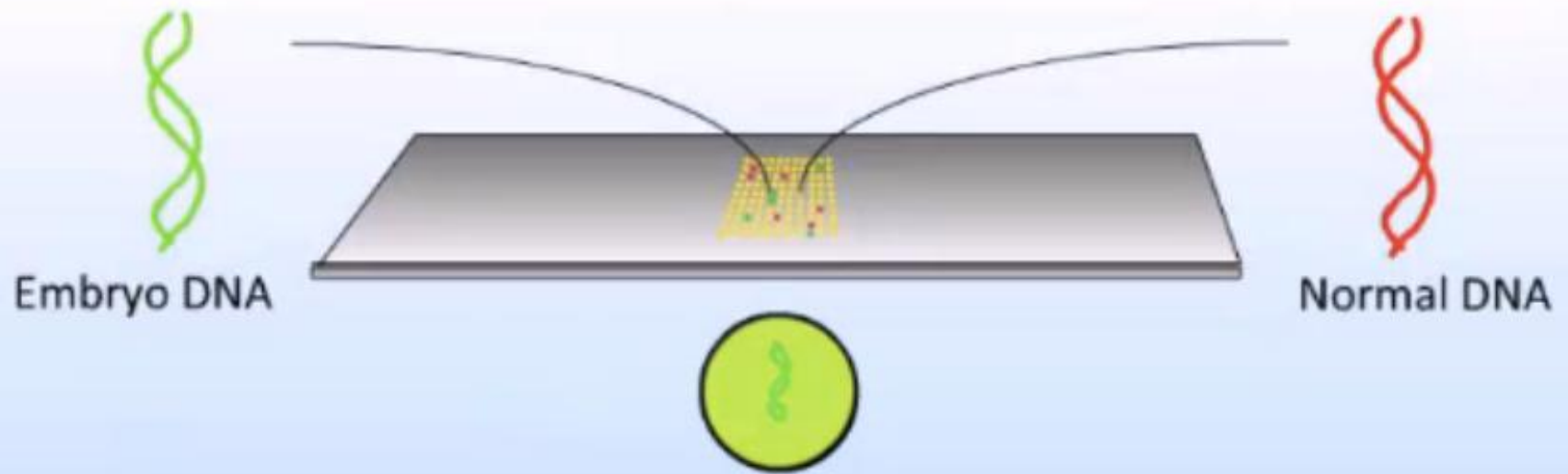
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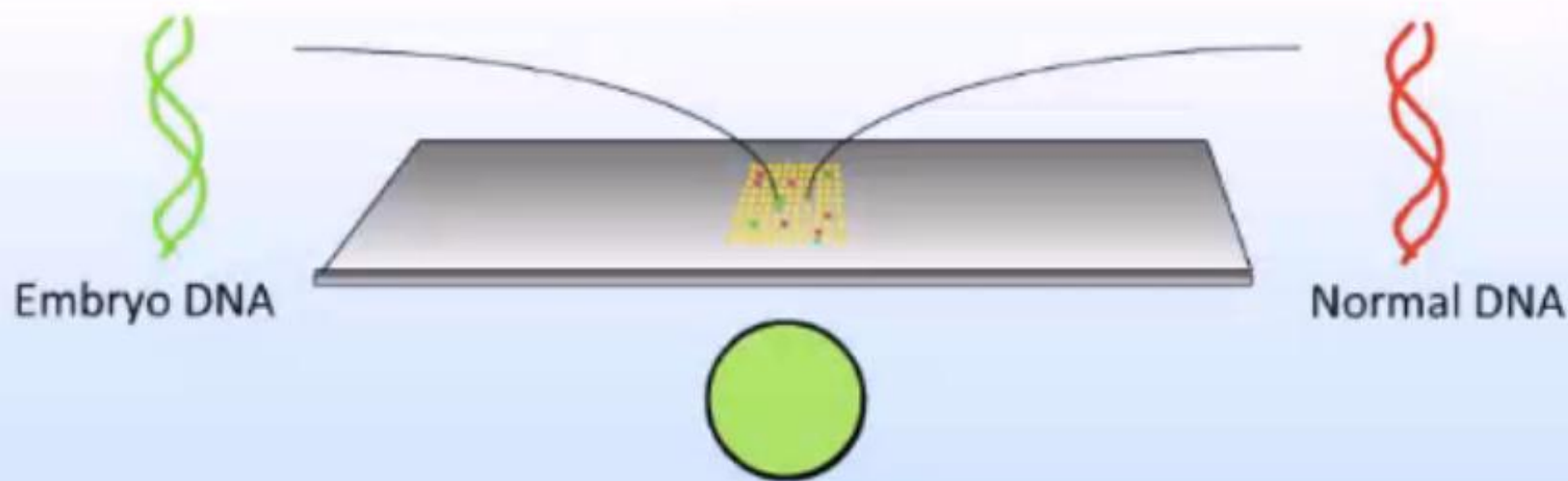
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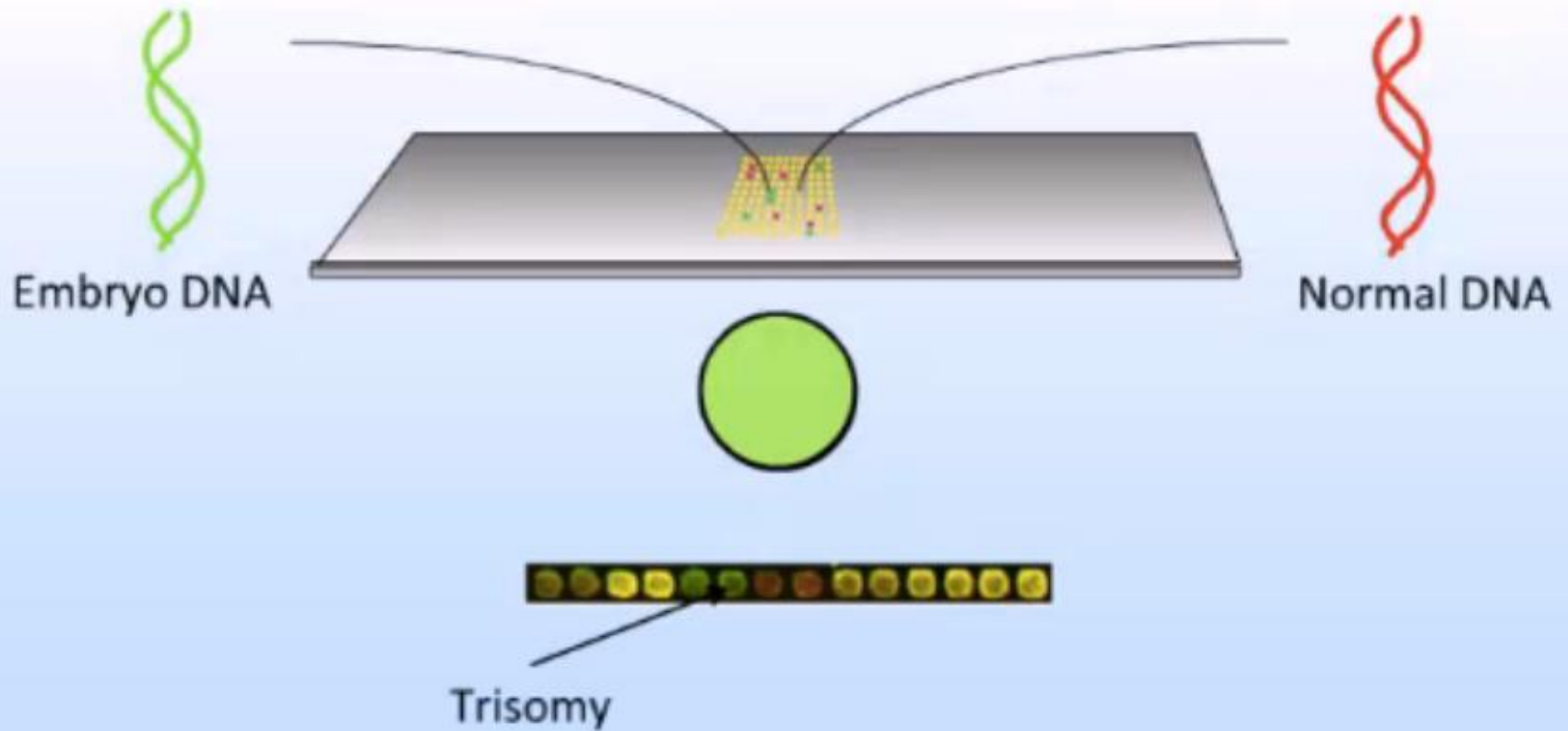
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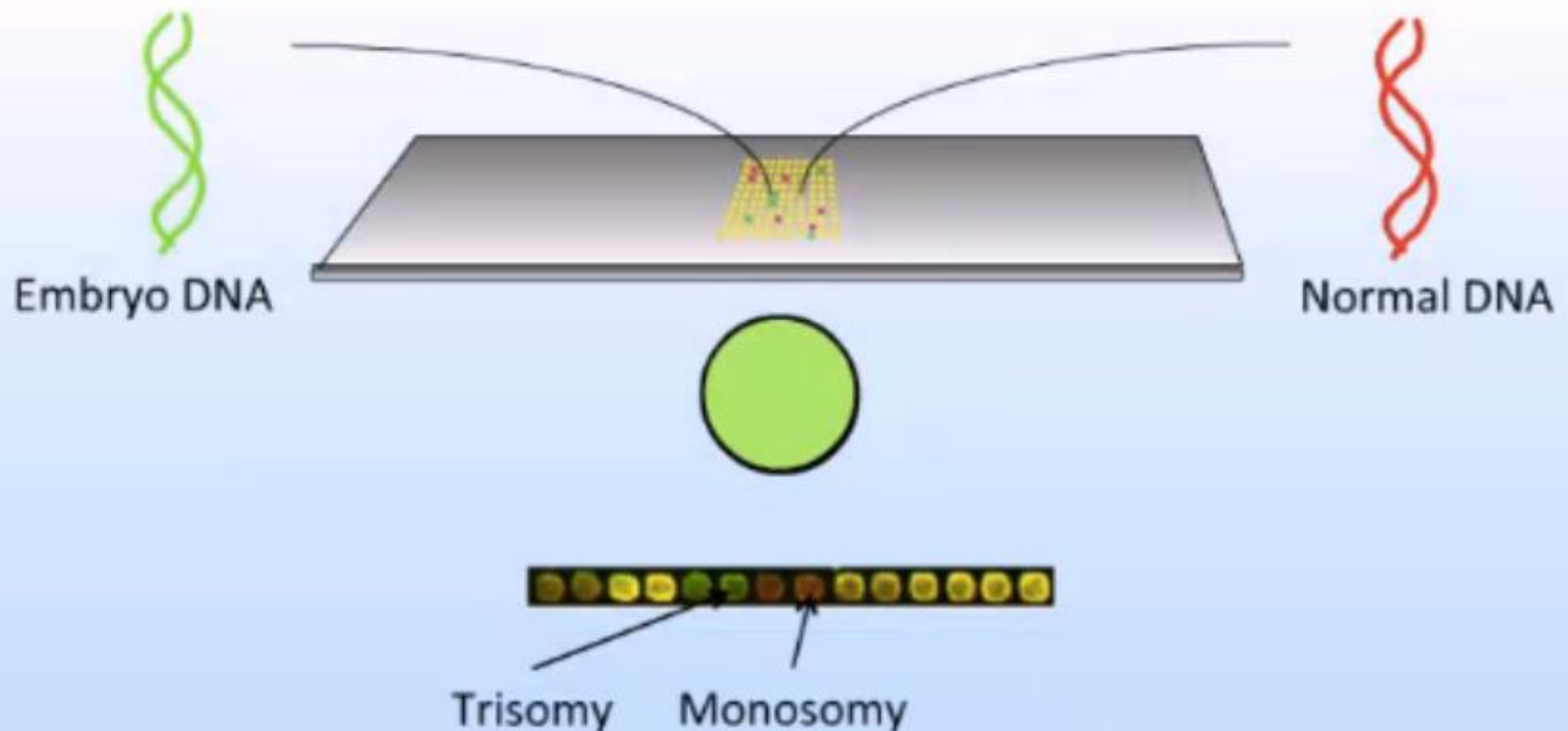
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