



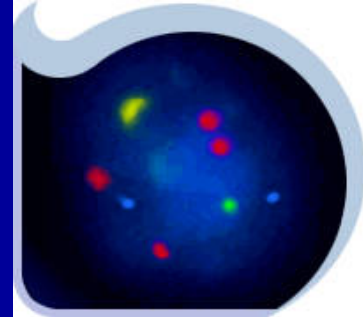
# Preimplantation Genetic Diagnosis

**Arjumand Warsy**

- **Preimplantation genetic diagnosis**

comprises a series of techniques used to:

- **diagnose** and
- **exclude genetic abnormalities** of embryos, **prior to transfer of the embryo or embryos** back to the uterus.

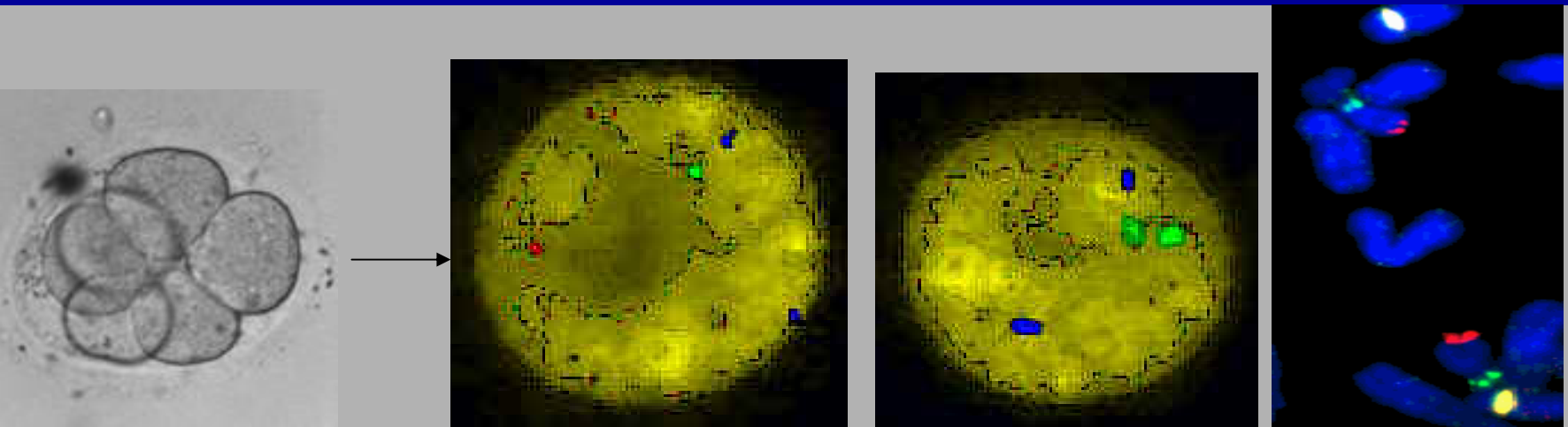


- The technique involves single cell biopsy of one or two cells from an embryo at the eight-cell stage.



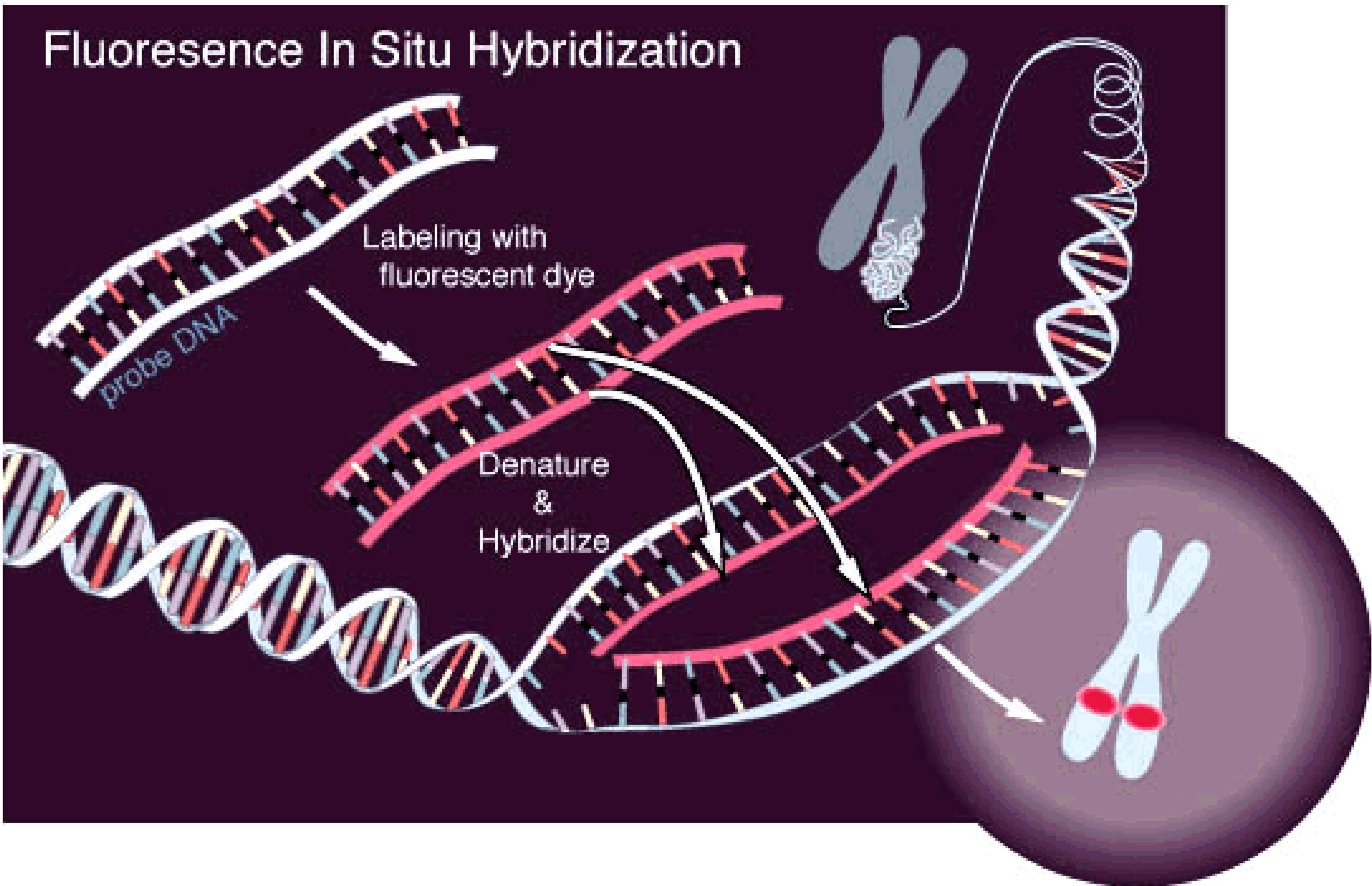
# PGD for sex-linked diseases

- Involves analysis of cells to determine the sex of the embryo by 'fluorescent in situ hybridization (FISH)'.
- FISH technique identifies chromosomes 13, 18, 21, X and Y and to look for other specific whole chromosome problems.



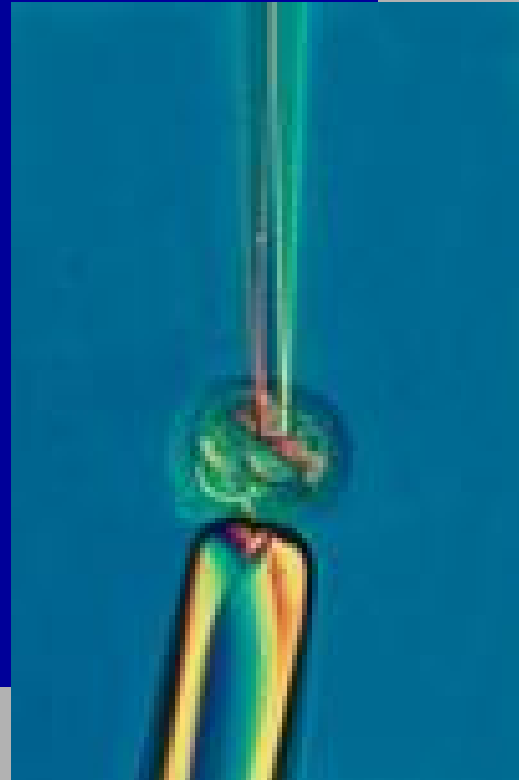
# Fluorescent In Situ Hybridization

## Fluorescence In Situ Hybridization

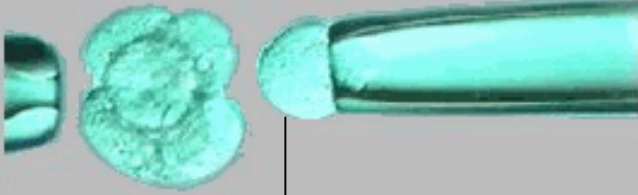


# Single cell PCR and PGD

- Single cell analysis by PGD is used for couples who are carriers of different genetic conditions such as:
  - alpha-1-antitrypsin deficiency,
  - cystic fibrosis
  - muscular dystrophy and
  - huntington's chorea
  - sickle cell disease
  - beta thalassaemia



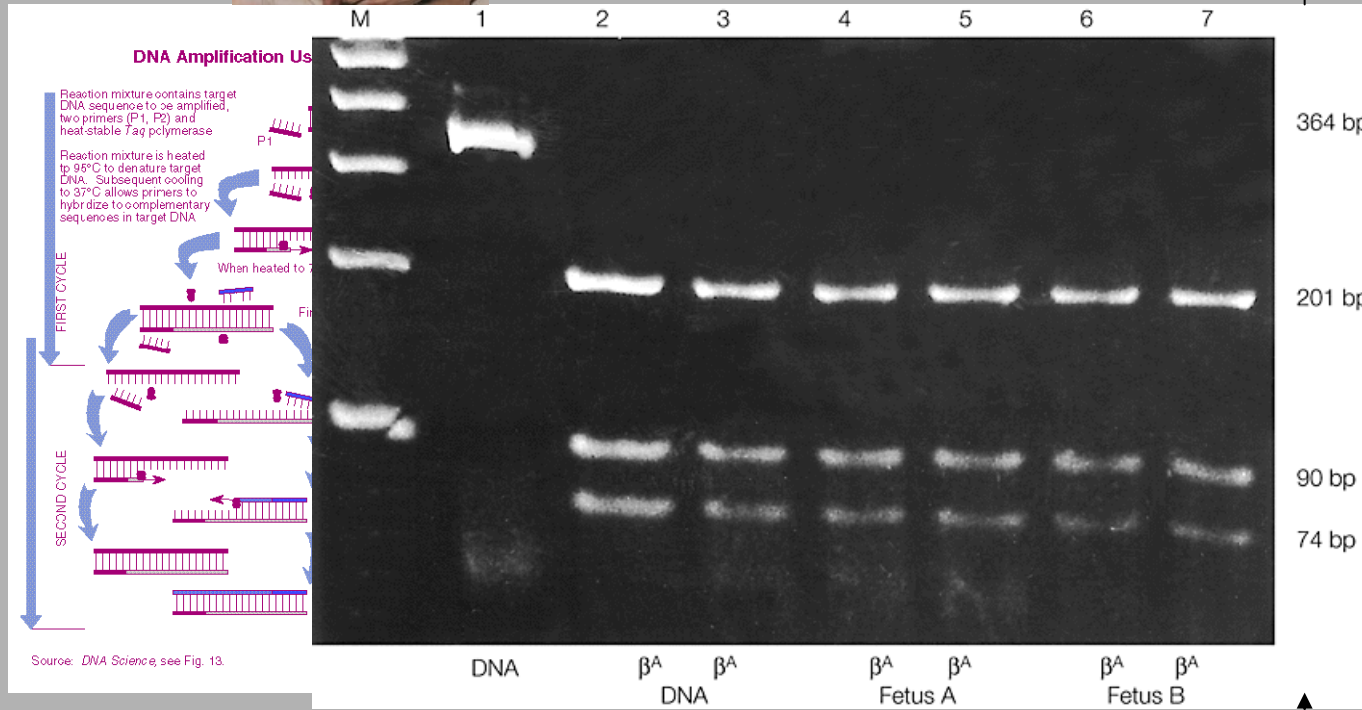
# Single cell analysis by PCR



Implantation in uterus

DNA

PCR amplification



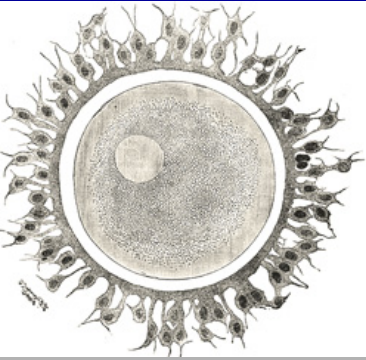
PCR product

Electrophoresis

# IVF and PGD

Induction of ovulation using medications to increase numbers of eggs developed

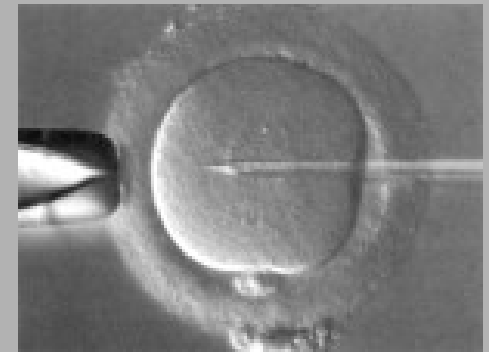
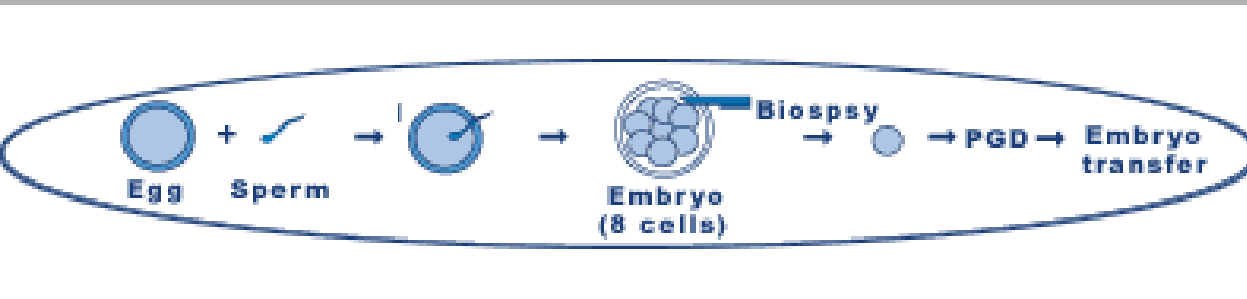
Retrieval of the eggs at an appropriate stage



Fertilization with the sperm and transfer to culture disc

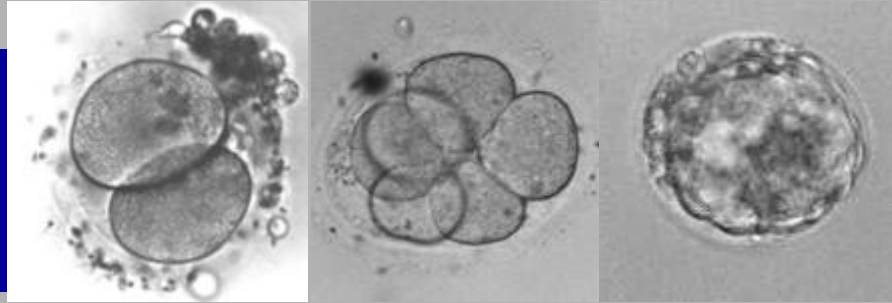
IVF

Intracytoplasmic sperm injection (ICSI)

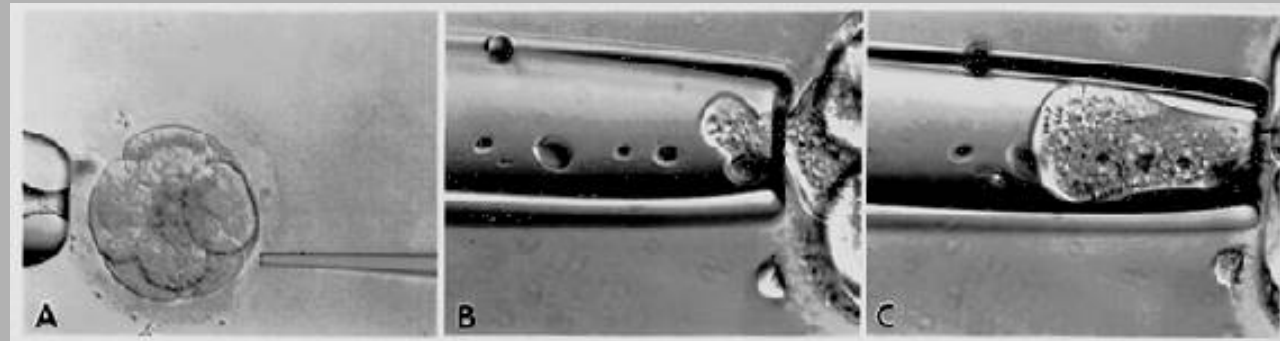


# IVF and PGD (contd..)

**Culturing of the pre-embryo to blastocyst stage**



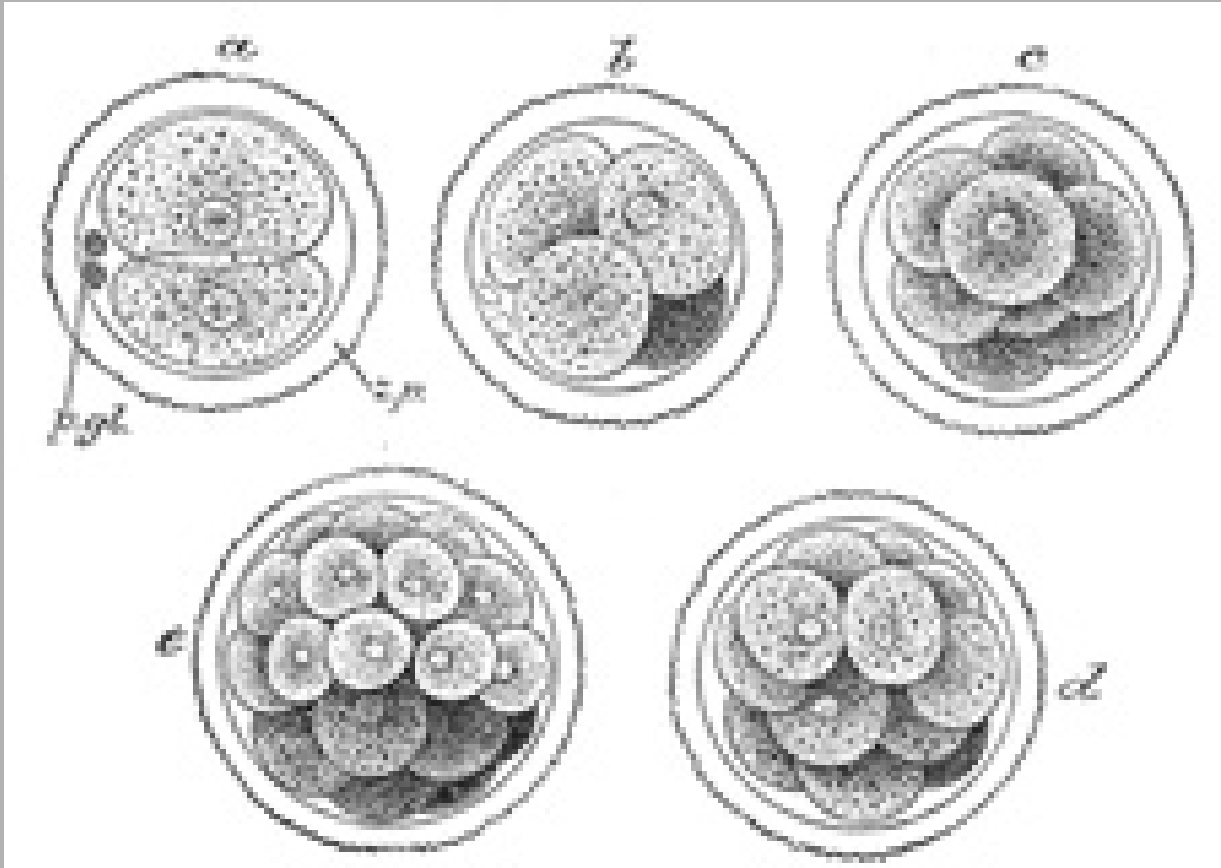
**Removal of single cell for diagnosis**



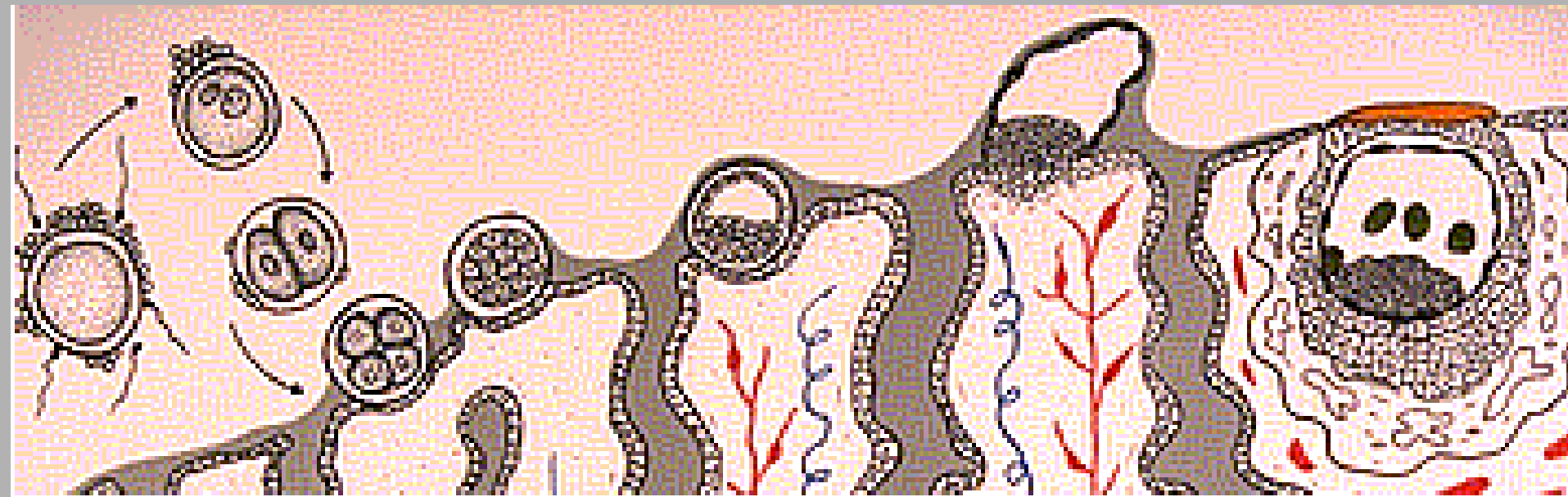
**If normal, transfer to uterus.**

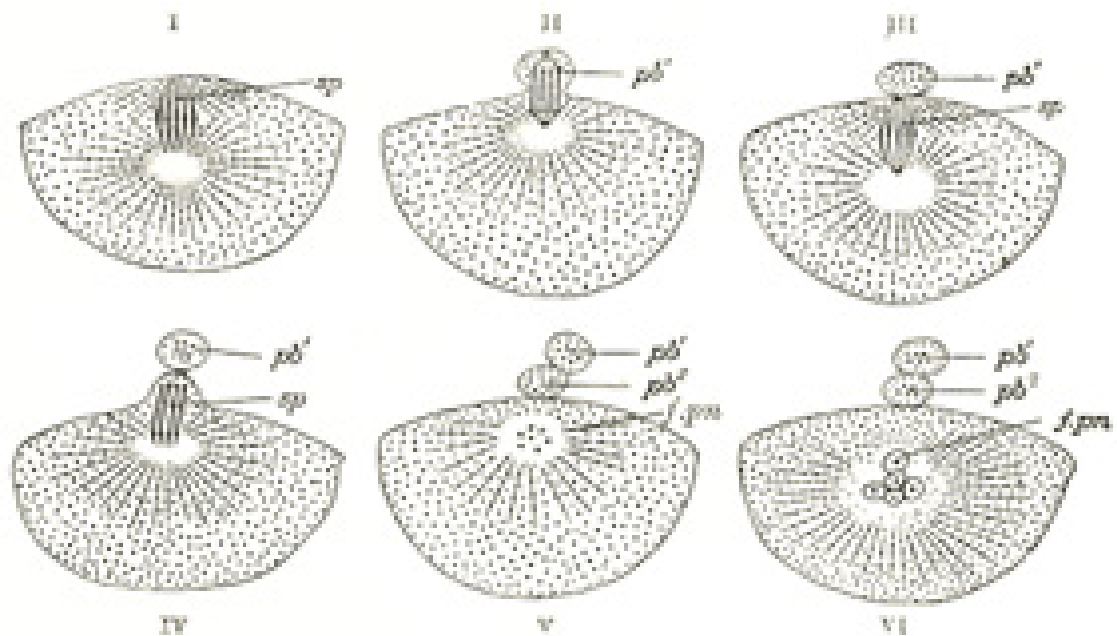
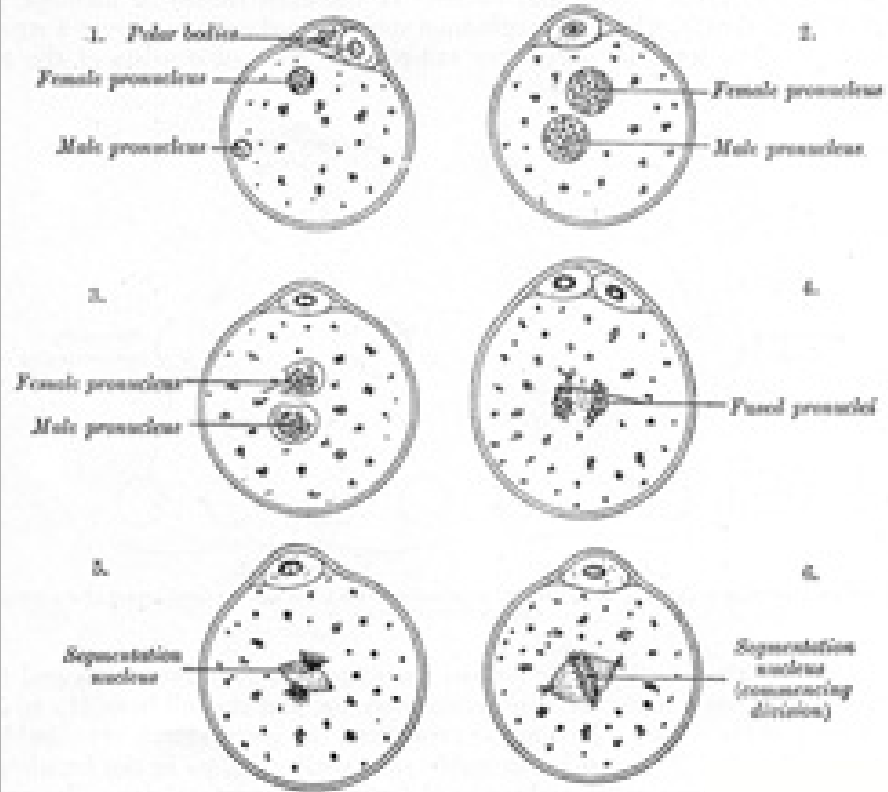


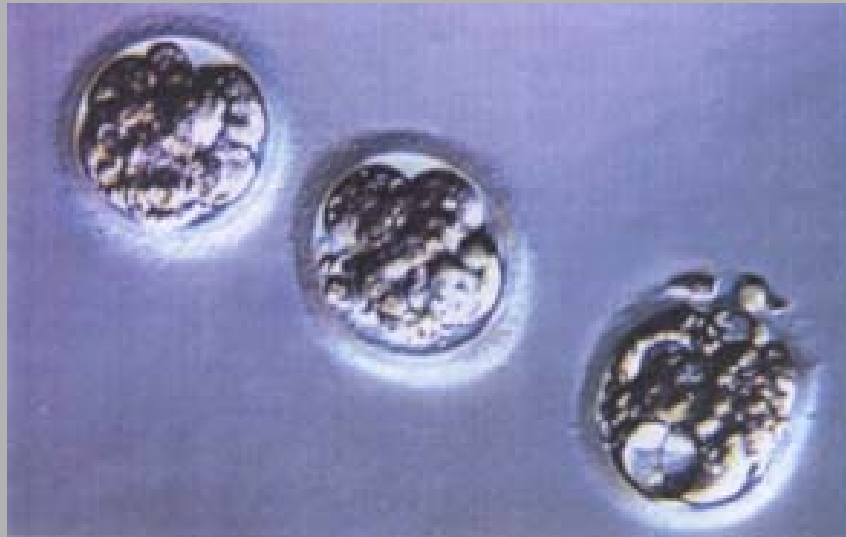


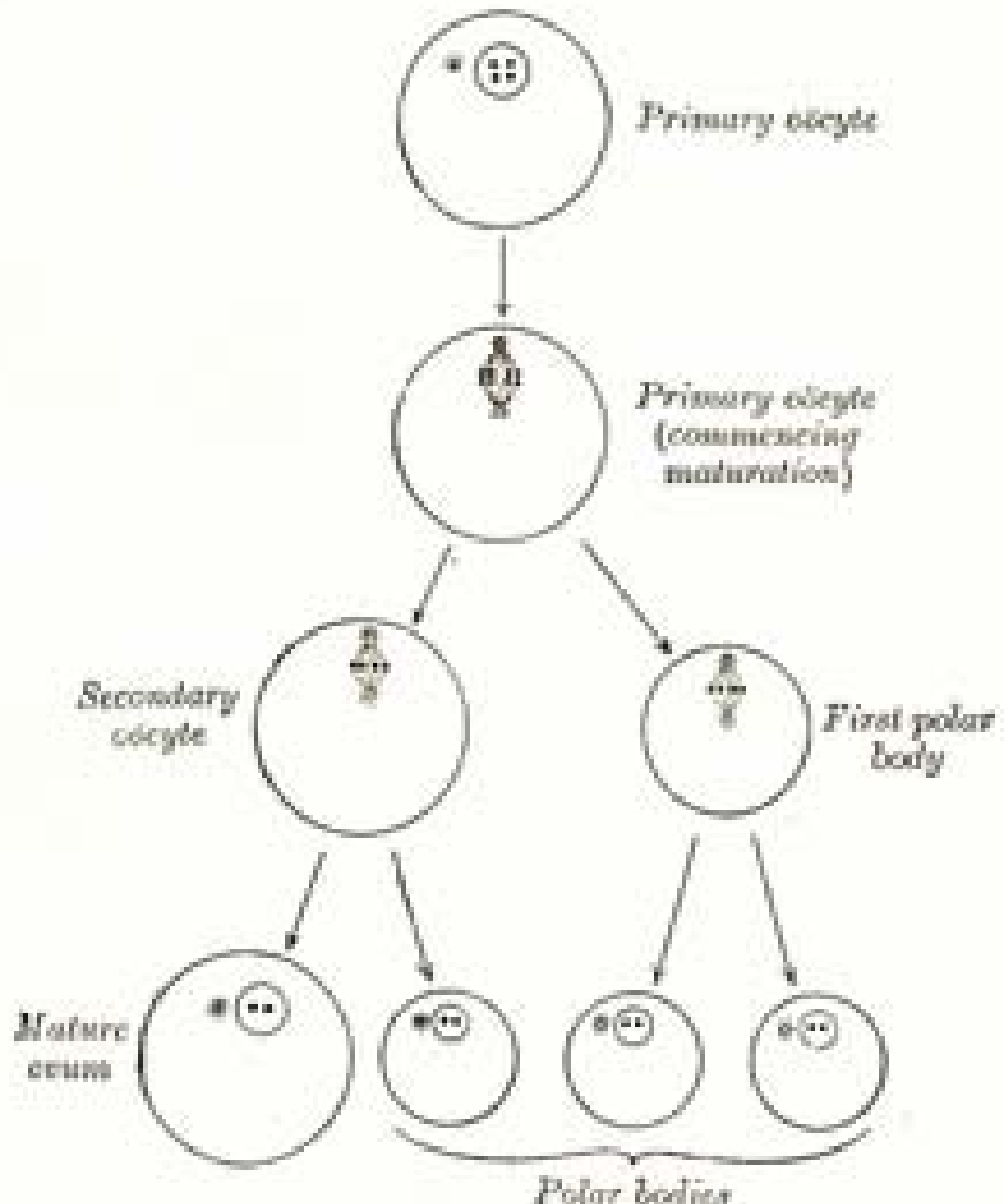


# In Vitro Fertilization- from insemination through embryo transfer to implantation









# PGD of sickle cell anaemia

