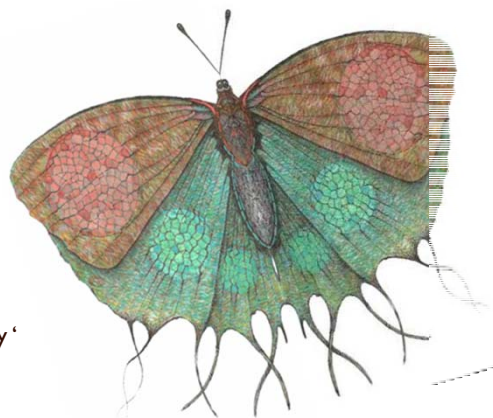


Pre-implantation Genetic Diagnosis (PGD)

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Westmead Hospital
'Genea – World Leaders in Fertility'
(formerly Sydney IVF)
October 2011



Pre-implantation genetic diagnosis (PGD)



- When a serious genetic disorder affects a person or a family member
- Combine IVF with genetic testing of embryos prior to transfer to the uterus
- Choice for family planning
- Alternative to prenatal genetic testing on an ongoing pregnancy

History



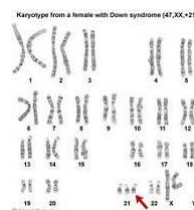
- 1967 - successful sexing of rabbit blastocysts
- 1978 - 1st IVF child born
- 1980s - human IVF fully developed
- 1990 first births from PGD



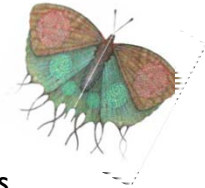
Indications for PGD?



- Specific condition where a mutation/gene is known
 - eg Haemophilia, cystic fibrosis
- Chromosome rearrangement
- Screen for abn chromosome number
 - eg Down syndrome
- Cancer predisposition
- Gender selection
- Non-disclosure eg HD
- HLA matching



Genea - PGD 'work ups' to date



- Total work ups for single gene disorders = 547

Cystic fibrosis = 13%

Huntington disease = 10%

Fragile X = 5%

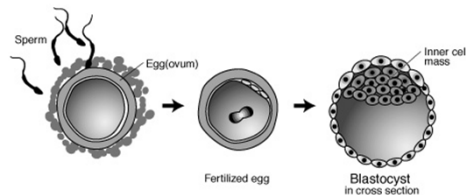
Neuromuscular Disorders = 16%

Haematological Conditions = 11%

eg. Thalassemia, Haemophilia

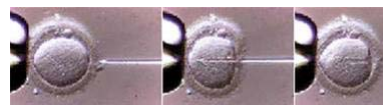
Process

- IVF




- ICSI (intracytoplasmic sperm injection)


- Single sperm injected into each egg
- To avoid the risk of contamination




Embryo development



Oocyte




Zygote




4 cell


Day 3Day 5



8 cell



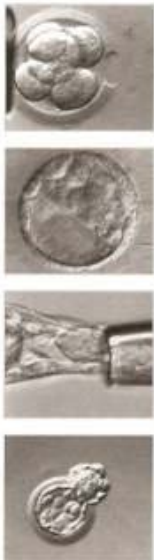
Morula




Blastocyst

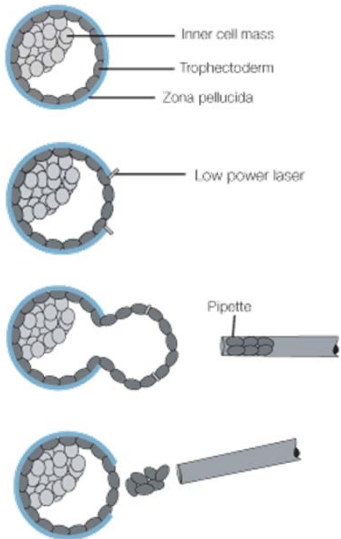
Biopsy at blastocyst stage

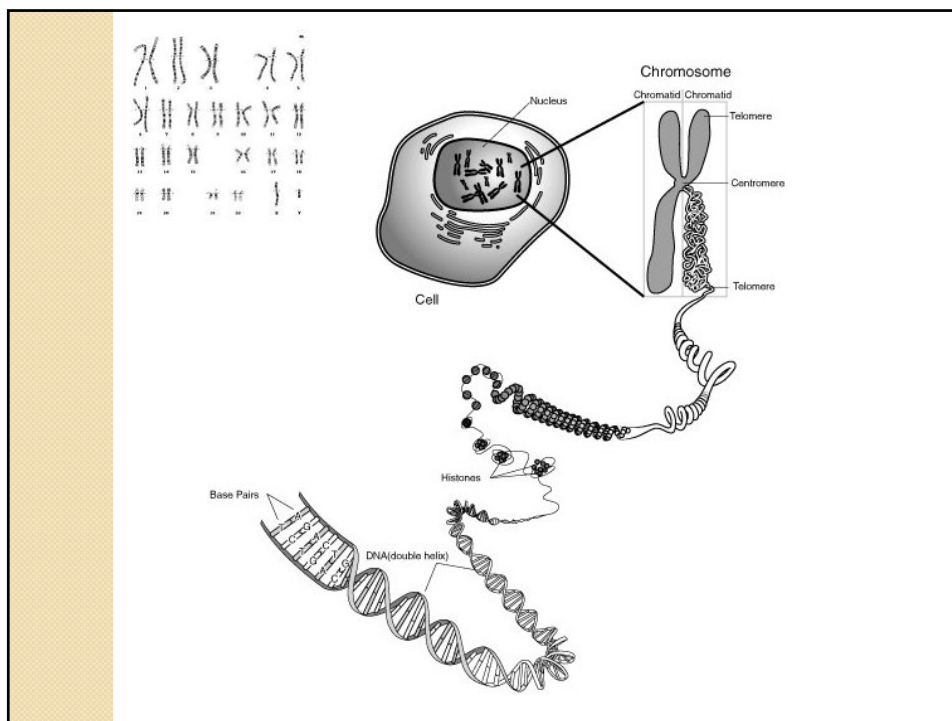
Day 5/6 post fertilisation





Biopsy.mpg





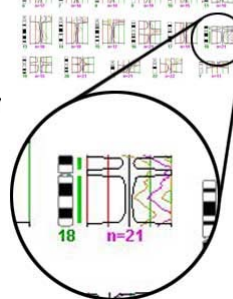
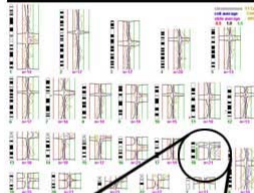
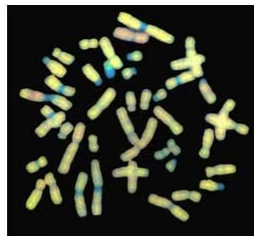
Genetic analysis



- Can only be done when the specific genetic mutation is known
 - or we are certain of the gene involved
- Usually have 20,000 - 100,000 cells
 - here only a few cells
- Molecular analysis using PCR
 - use two methods
 - Linkage +/- direct mutation
 - >99% accurate

Genetic analysis

- CGH (comparative genom hybridisation)
- Detects abnormalities in t number of all the chromosomes
 - eg. Down syndrome
 - Improves chance of an embri implanting

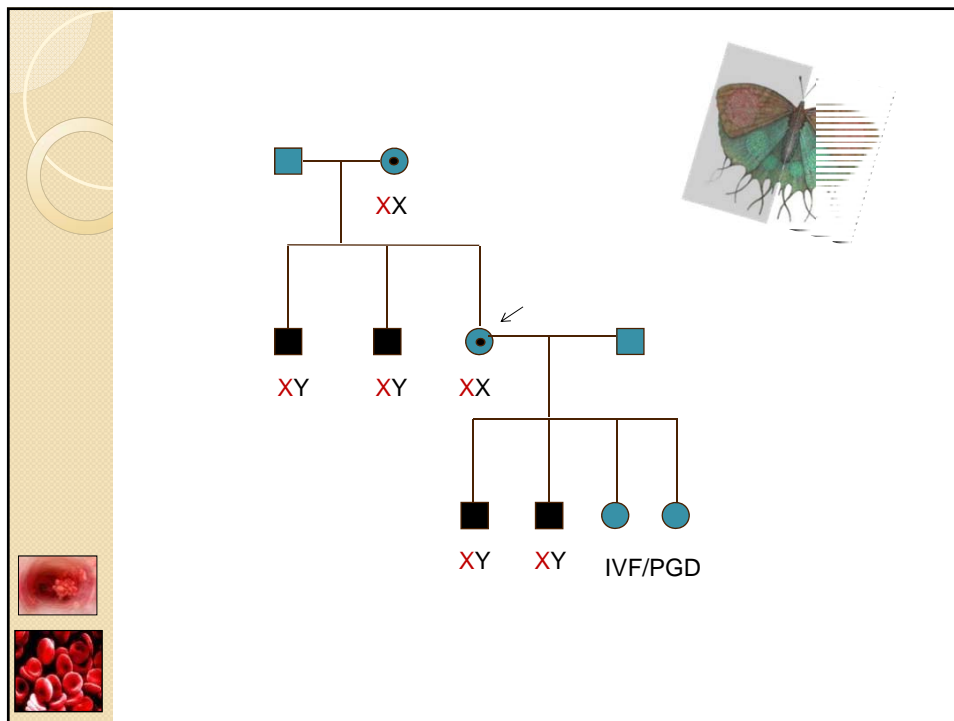
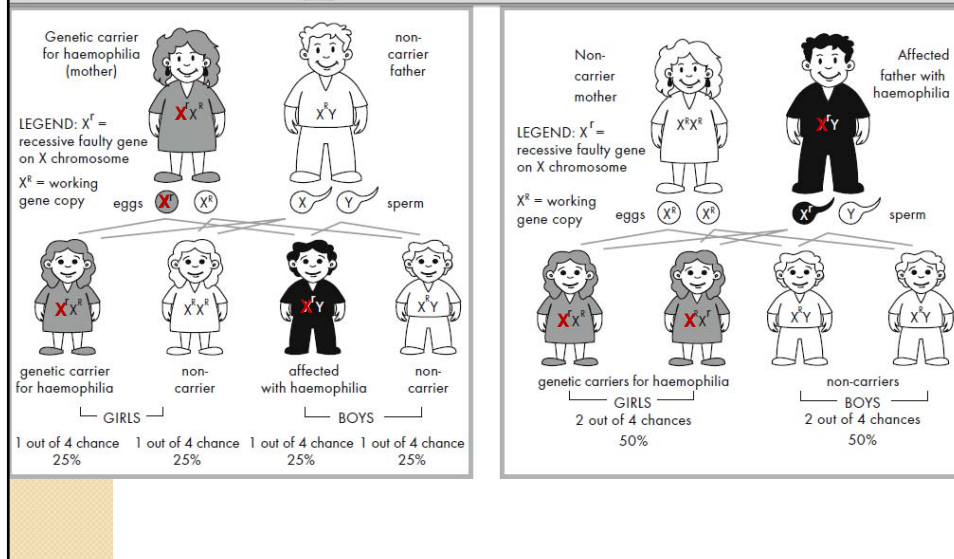


Pros and cons

- Risks of
 - IVF process for woman
 - IVF for baby
 - biopsy process
 - getting it wrong
- Alternatives – balance
- Chance of success?
 - Maternal age



Genetics of Haemophilia X-linked recessive inheritance



PGD Assistance



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Sydney IVF is now reborn as **GENEA**

Why the Italian outbreak?
On 10 September 2011, the first day of spring, Sydney IVF will be reborn as Genea, World leaders in fertility.

You are here: Home / How we can help / Our Services / PGD Assistance Program

About the PGD Assistance Program

Genea is pleased to support the PGD Assistance Program. This Program has been created by generous donors to provide financial assistance to families in the community who would otherwise be unable to afford Preimplantation Genetic Diagnosis (PGD).

PGD is an advanced screening technique that can determine whether an embryo has a genetic condition, common examples include cystic fibrosis, Huntington disease or muscular dystrophy. Tests for over 100 different genetic conditions have been developed to date. More tests are being developed as required. PGD is most commonly used by couples who are aware of the possibility that their children risk inheriting a serious genetic condition.

The PGD Assistance Program provides couples with financial assistance to access Genea's PGD services.

Who is it open to?

8 PGD ASSISTANCE PROGRAM

Fitness for Fertility >

Pay Online Now >

- Our Services
 - Assisted Conception
 - Genetic Screening

Medicare rebate for expensive IVF treatment PGD sought by clinics

The Daily Telegraph
July 22, 2011
11:40PM

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