



Problem
Challenge
Need

...

Solution
Innovation
Invention

...

Tubal Infertility

In-vitro Fertilisation

Steptoe and Edwards

Birth after the reimplantation of a
human embryo.

Lancet 1978

**Louise Brown,
25. Juli 1978**



Lesley and John Brown



Patrick Steptoe, Robert Edwards

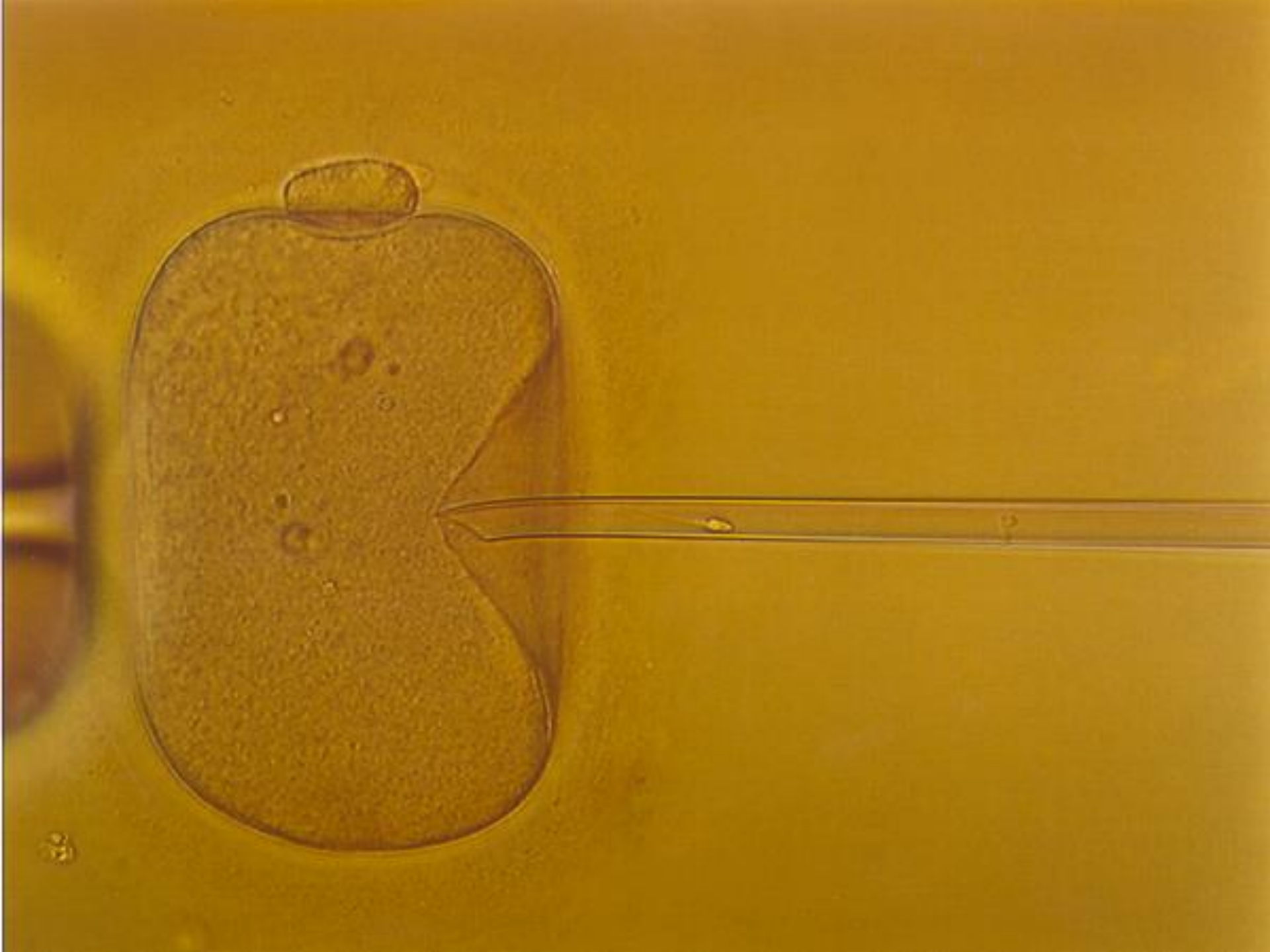
Male Infertility

Intracytoplasmic Sperm Injection (ICSI)

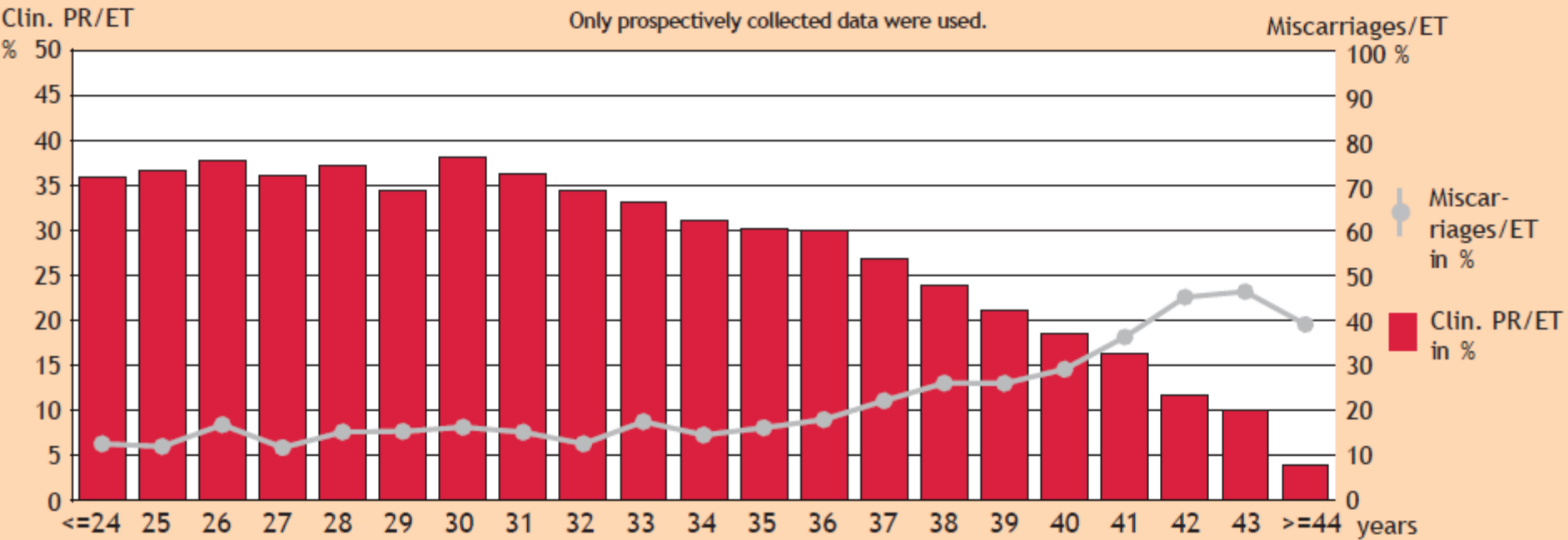
Palermo G

Pregnancies after intracytoplasmic injection of single spermatozoon into an oocyte.

Lancet 1992

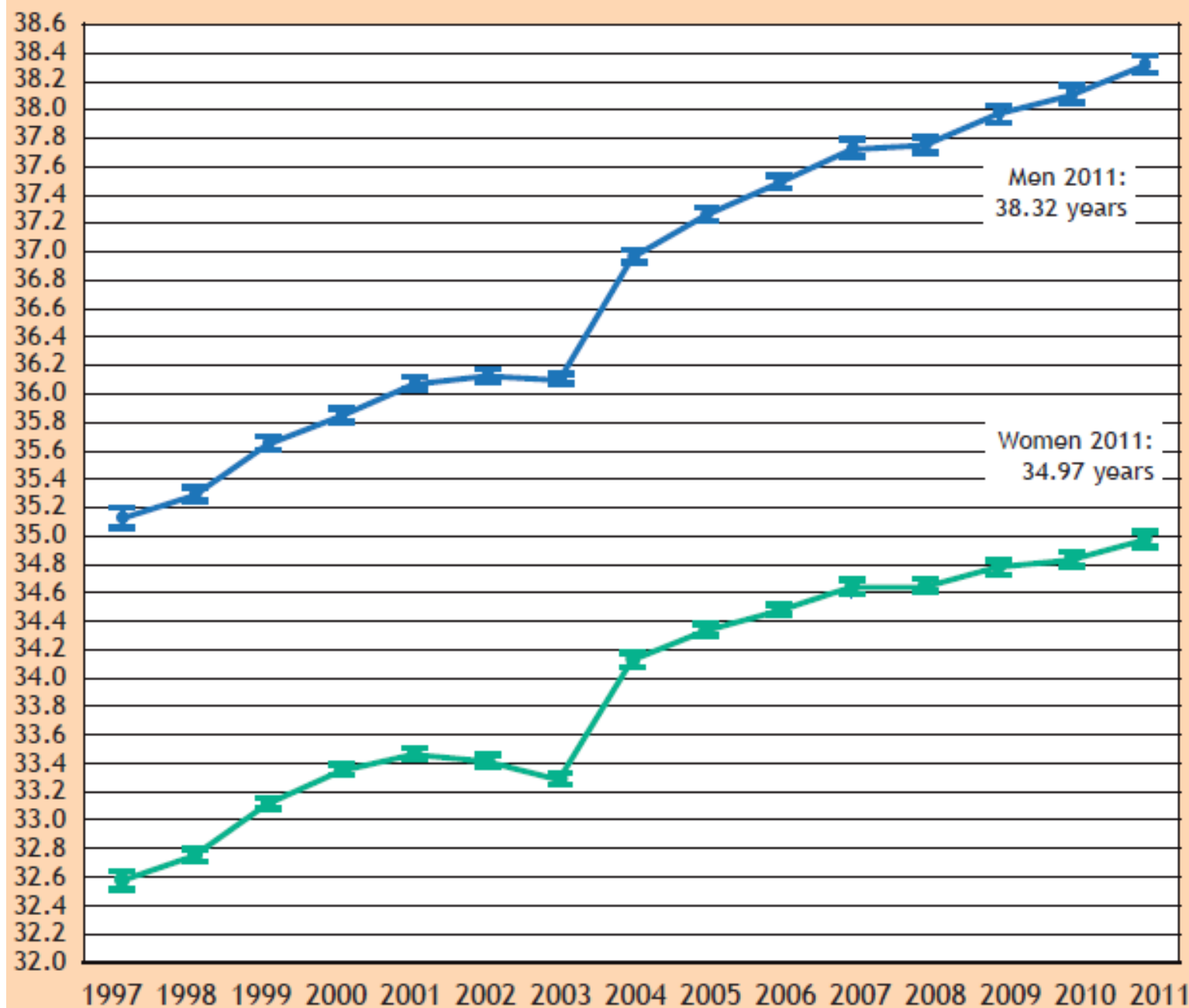


Advanced female age



Advancing female age

Age in years



Preimplantation genetic
screening (PGS) ?

P

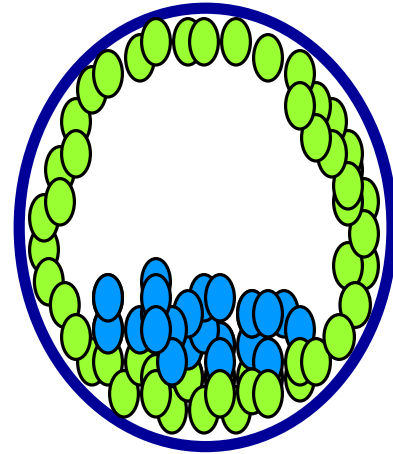
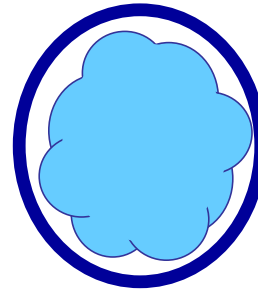
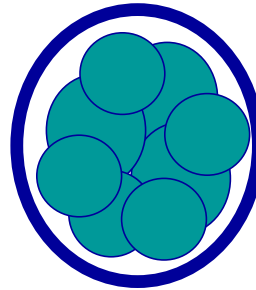
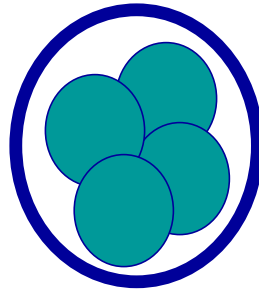
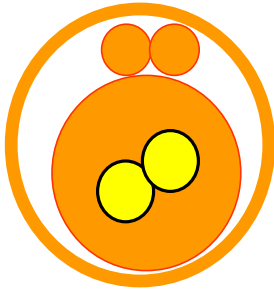
1

2

3

4

5



P

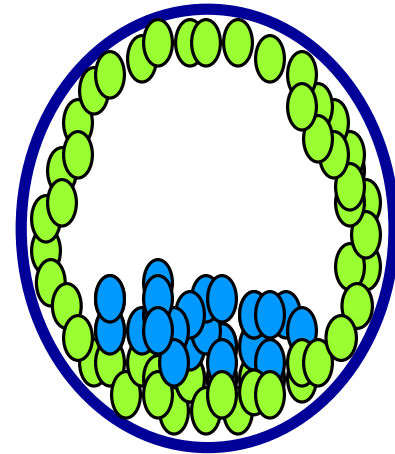
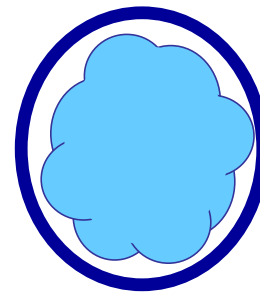
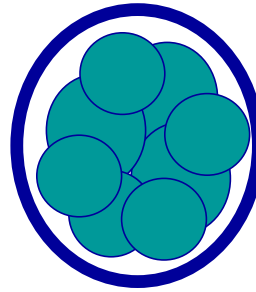
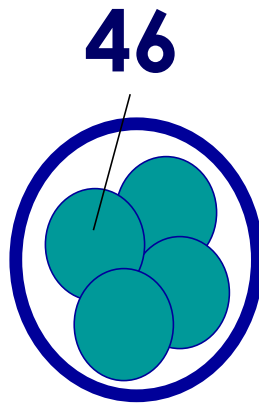
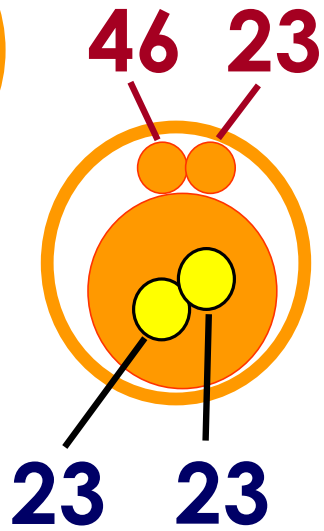
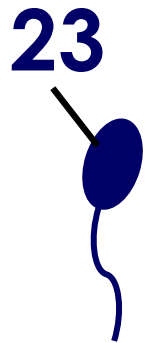
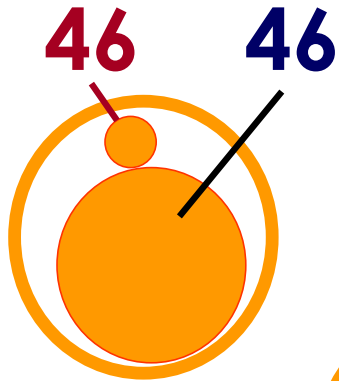
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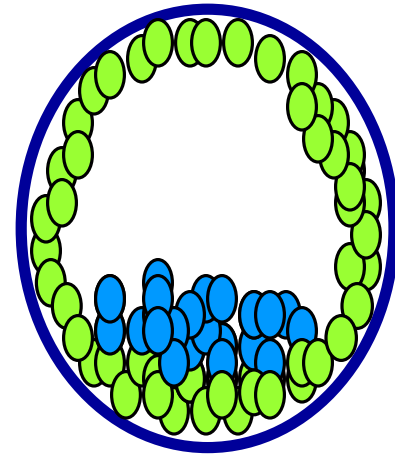
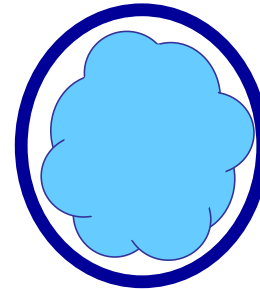
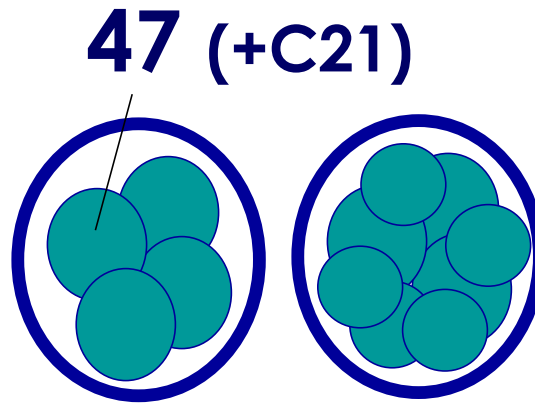
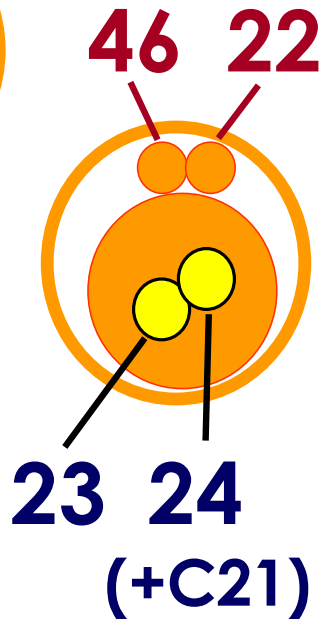
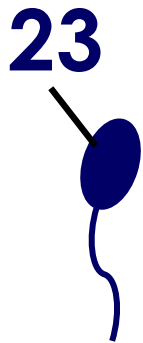
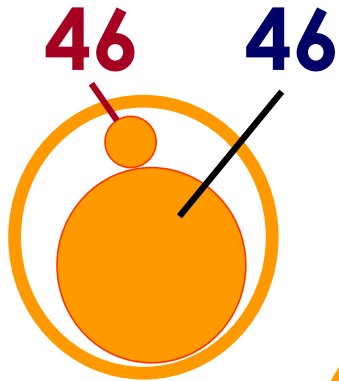
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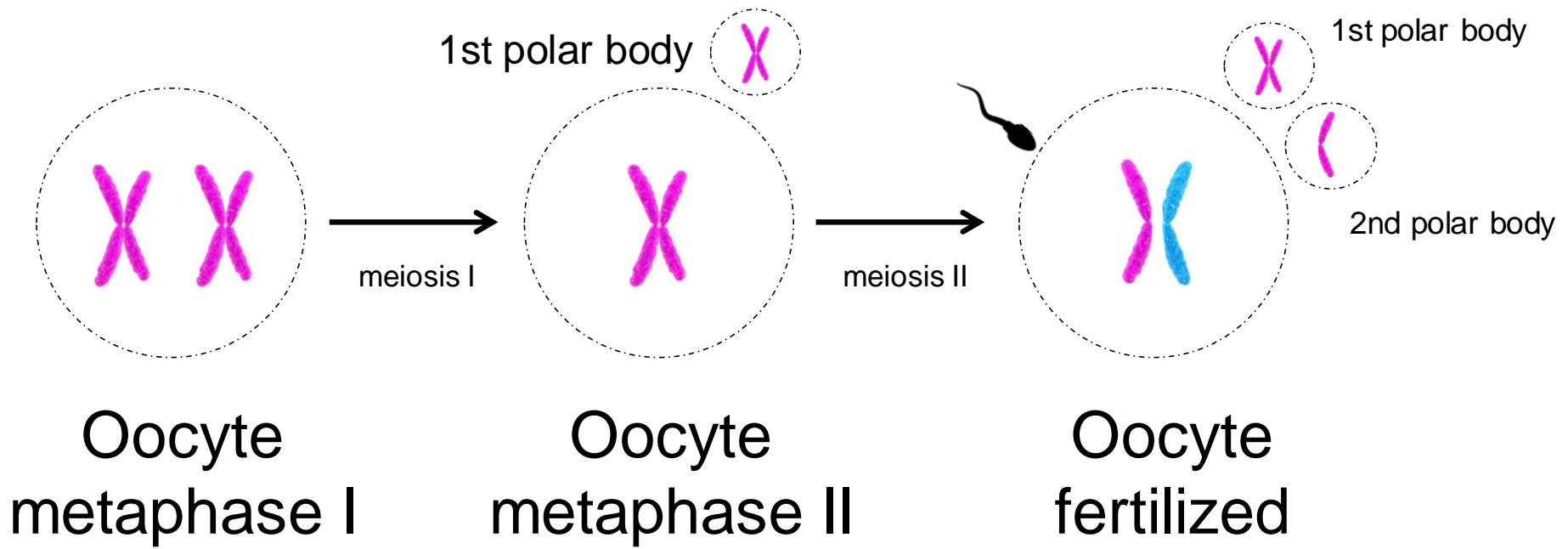
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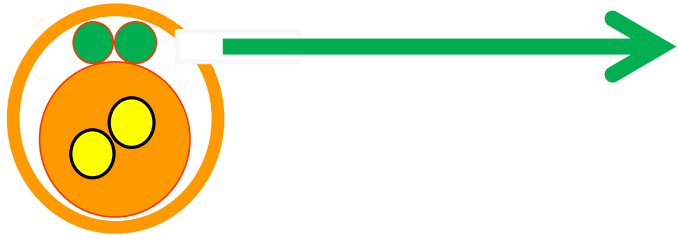
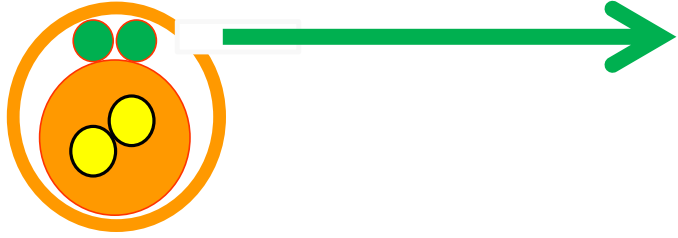
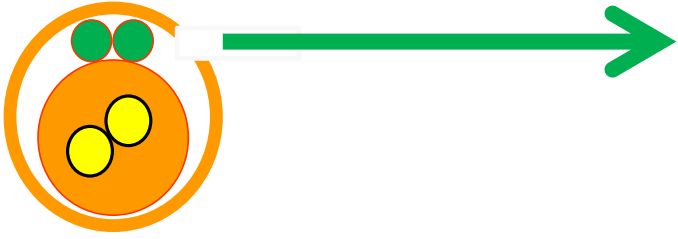


Verlinsky Y

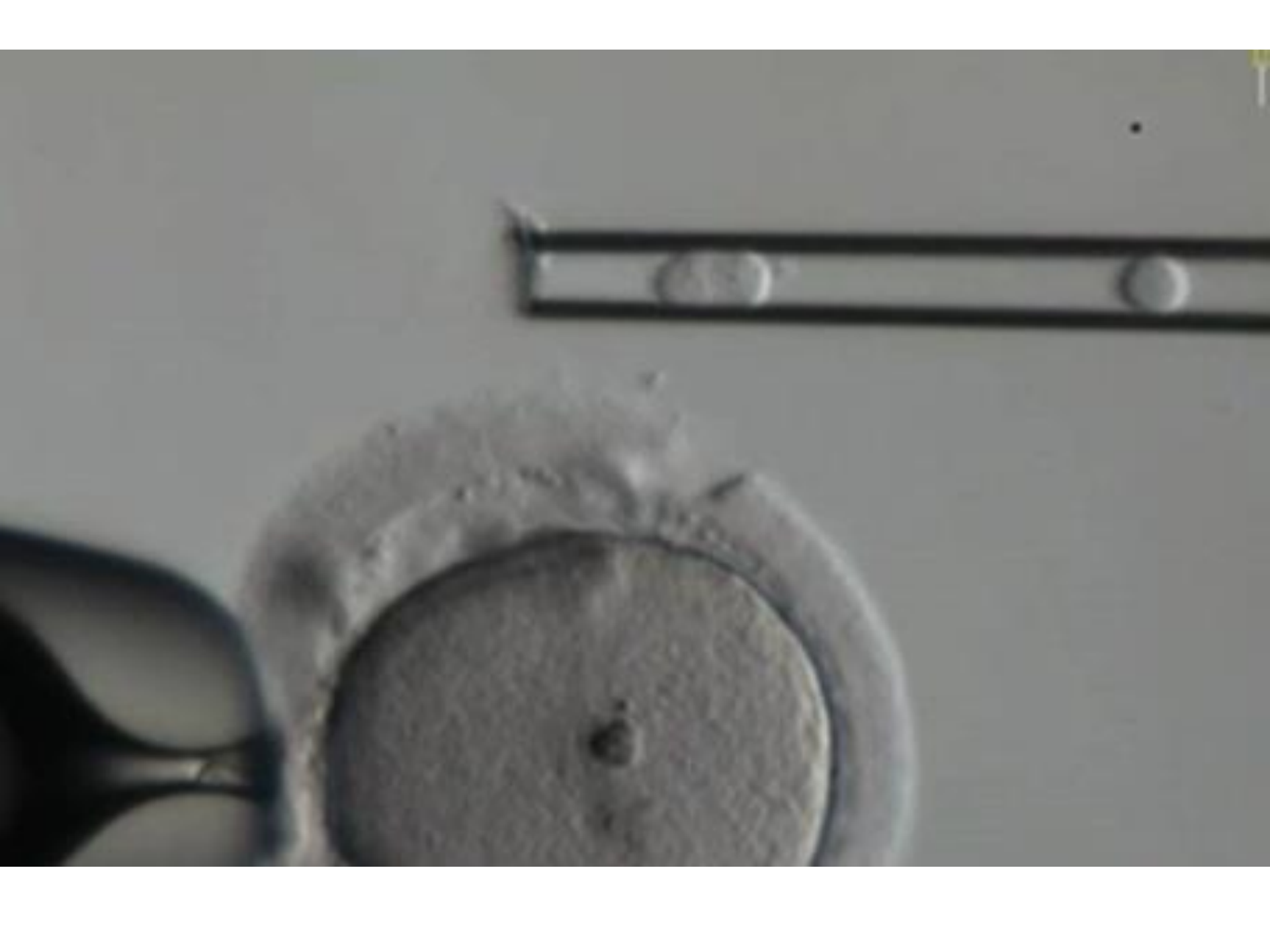
Ginsberg N, Lifchez A. et al. Analysis of the first polar body: preconception genetic diagnosis.

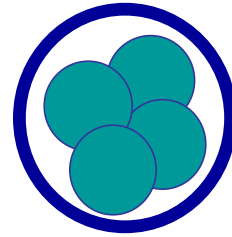
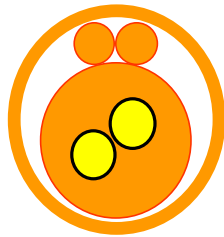
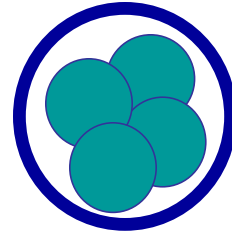
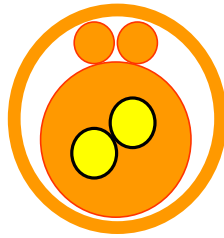
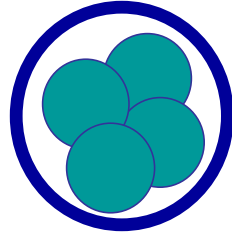
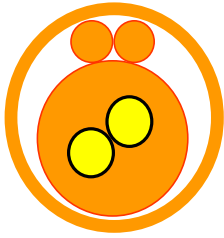
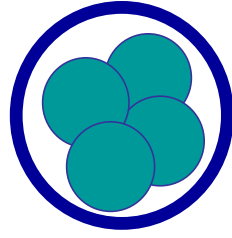
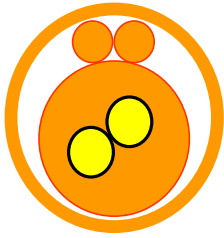
Hum Reprod. 1990







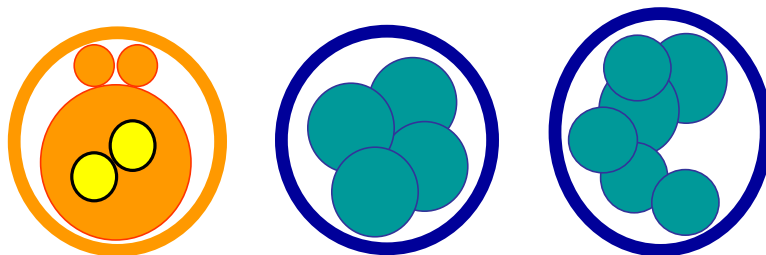
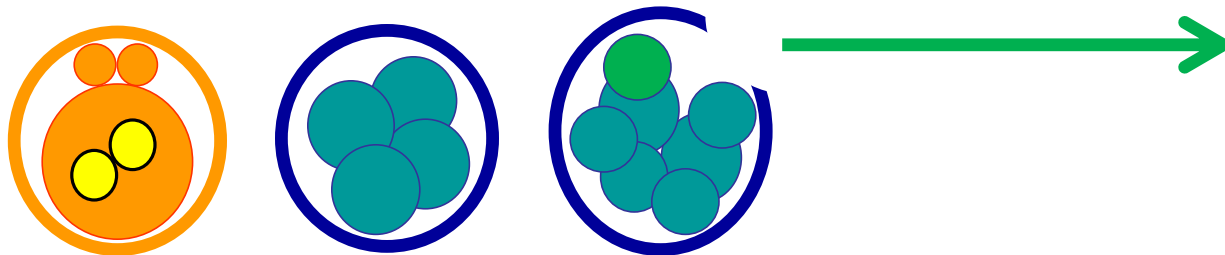
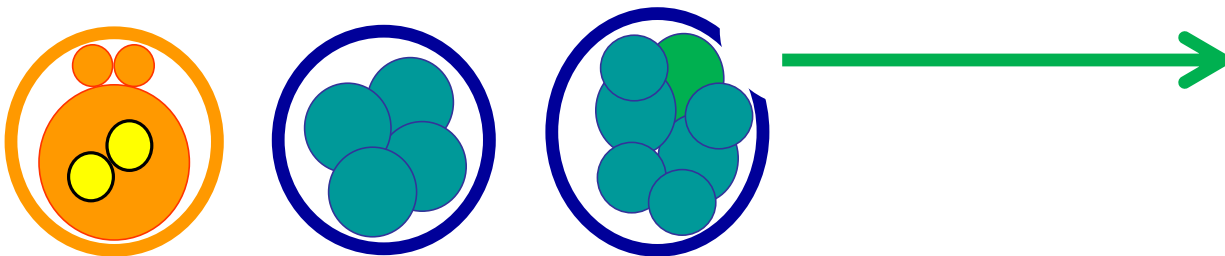
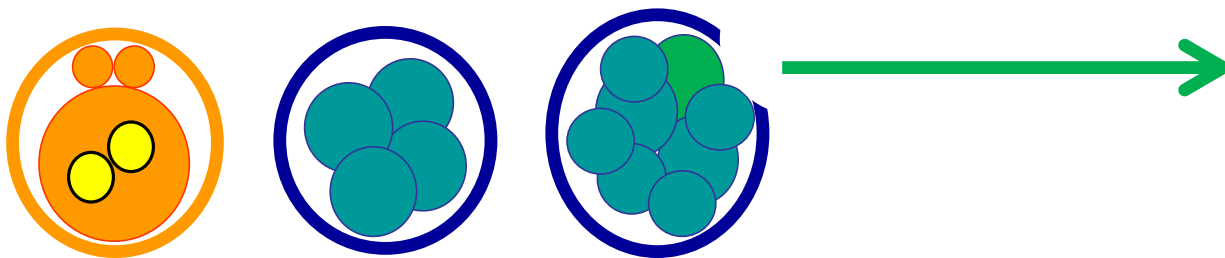




Handyside

Biopsy of human preimplantation
embryos and sexing by DNA
amplification.

Lancet 1989







b



Staessen

Comparison of blastocyst transfer with or without preimplantation genetic diagnosis for aneuploidy screening in couples with advanced maternal age: a prospective randomized controlled trial.

Hum Reprod 2004

Hardarson

Preimplantation genetic screening in women of advanced maternal age caused a **decrease** in clinical pregnancy rate: a randomized controlled trial.

Hum Reprod. 2008

Mastenbroek

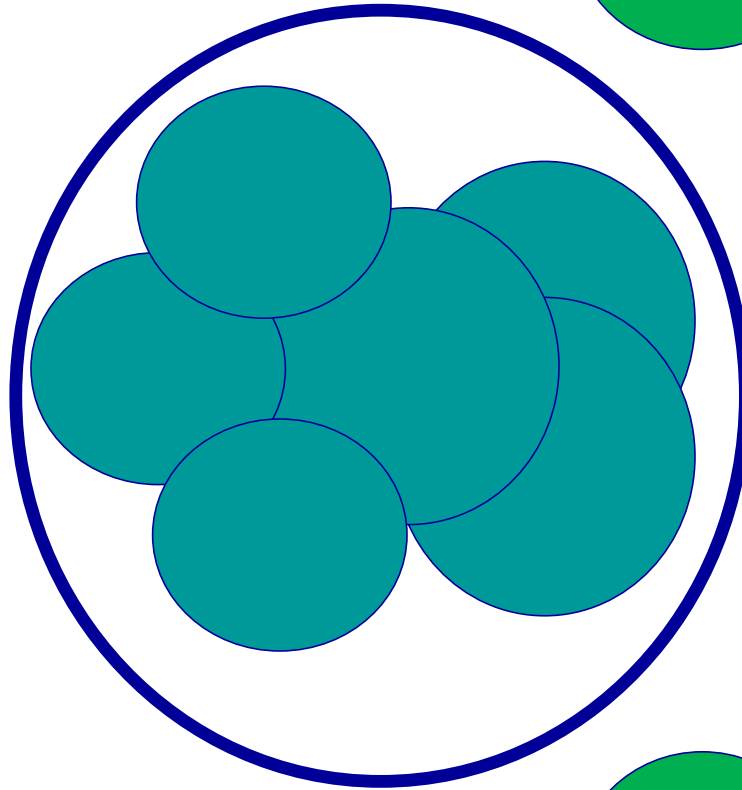
In vitro fertilization with preimplantation
genetic screening.

N Engl J Med. 2007

Evidence against the use of PGS for advanced maternal age patients when performing IVF.

1. Day 3 Blastomere Biopsy
2. Removal of 2 cells
3. FISH Analysis of only few chromosomes

1



2

De Vos

Impact of cleavage-stage embryo biopsy
in view of PGD on human blastocyst
implantation: a prospective cohort of
single embryo transfers.

Hum Reprod. 2009

At least 9 chromosomes should be tested:

chromosomes
analyzed

% Abnormal
fetuses detected

5

28 %

6

47 %

9

70 %

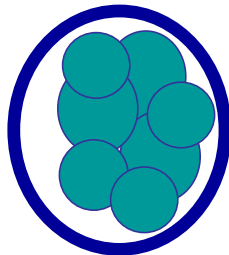
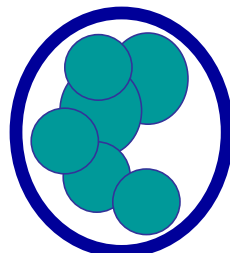
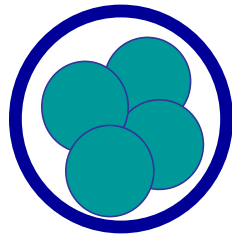
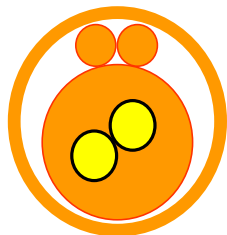
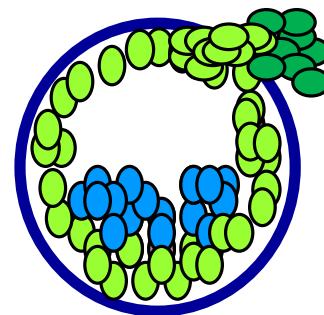
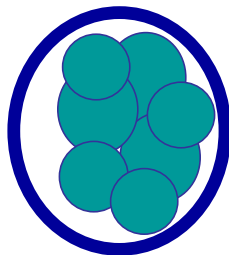
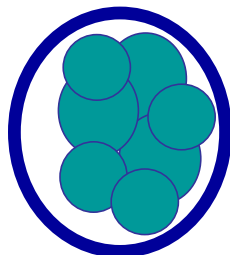
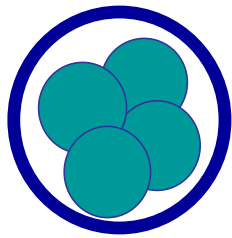
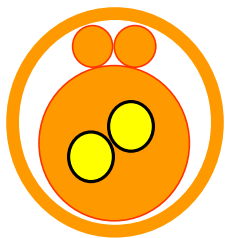
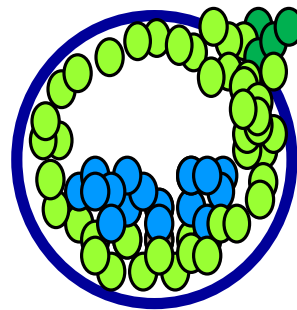
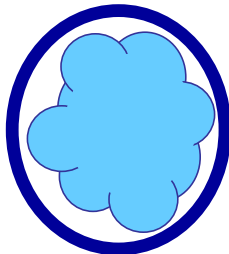
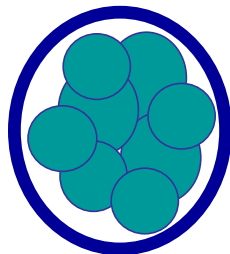
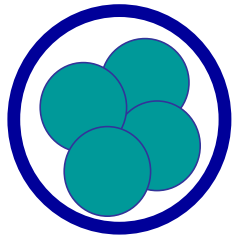
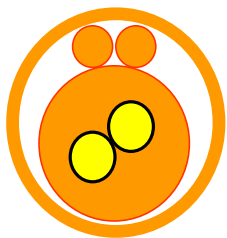
12

80 %

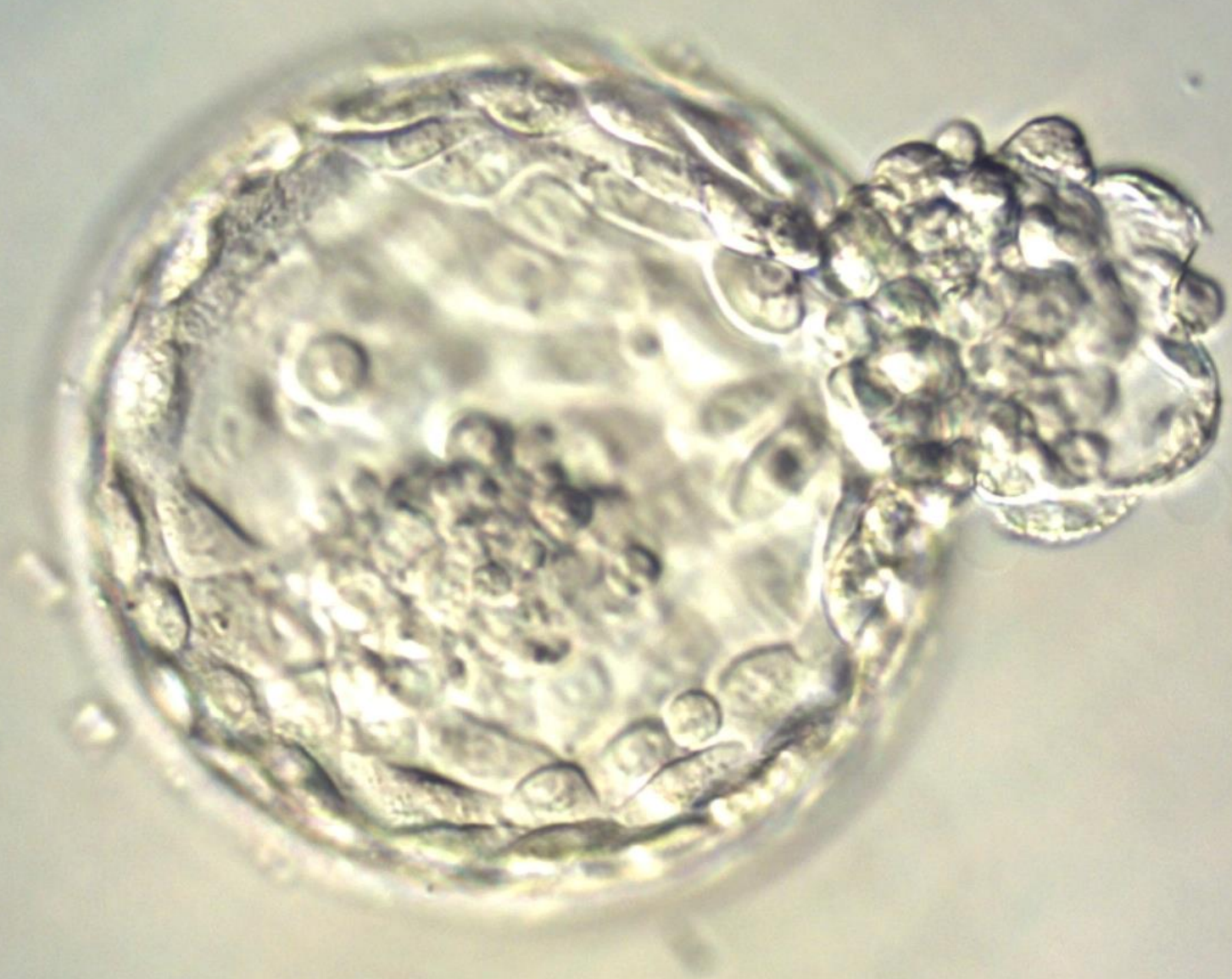
24

100 %

Blastocyst Culture
Vitrification
Biopsy









Benefits:

More DNA – more robust diagnosis

Reduces error rate

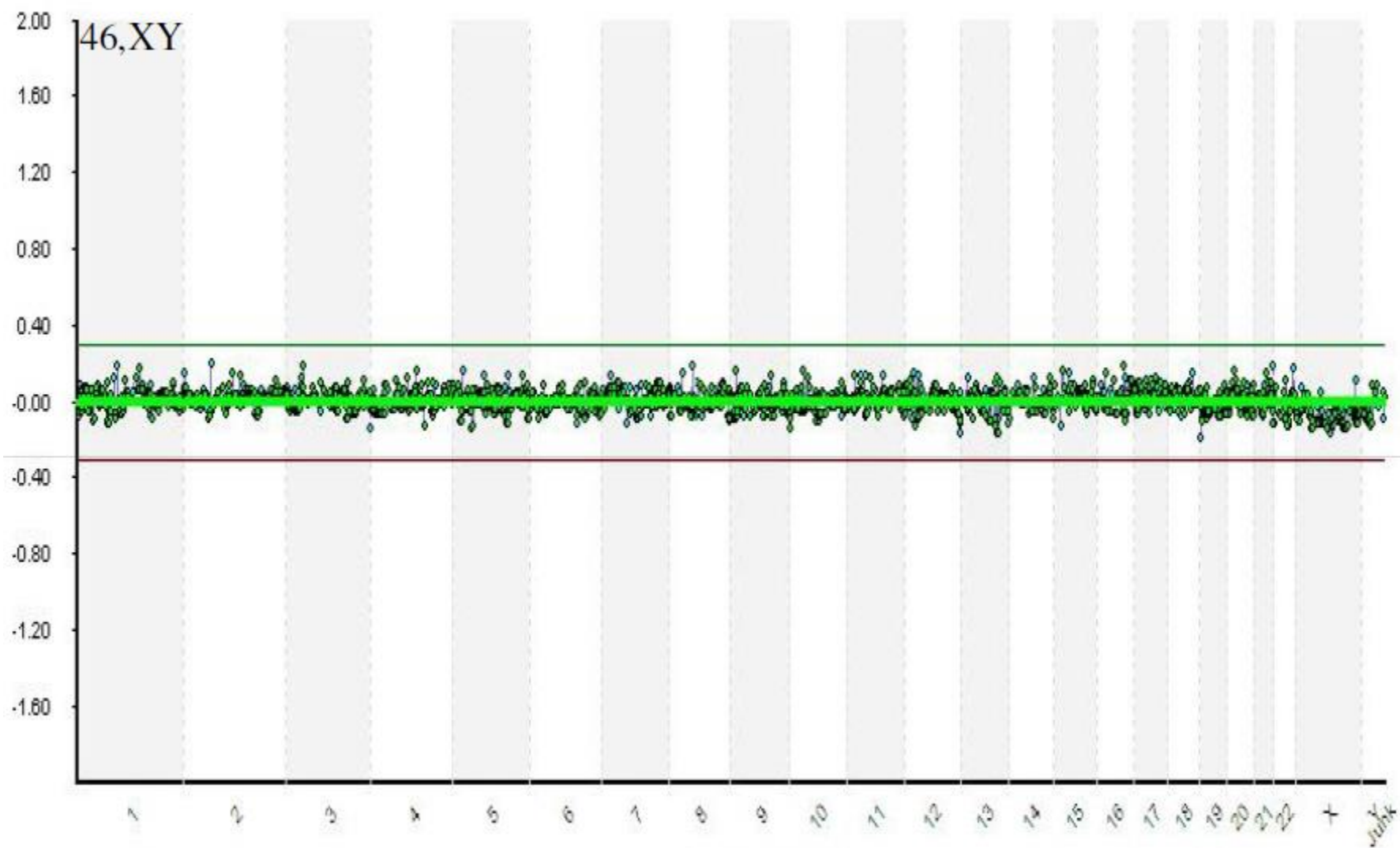
Reduced impact of embryo biopsy

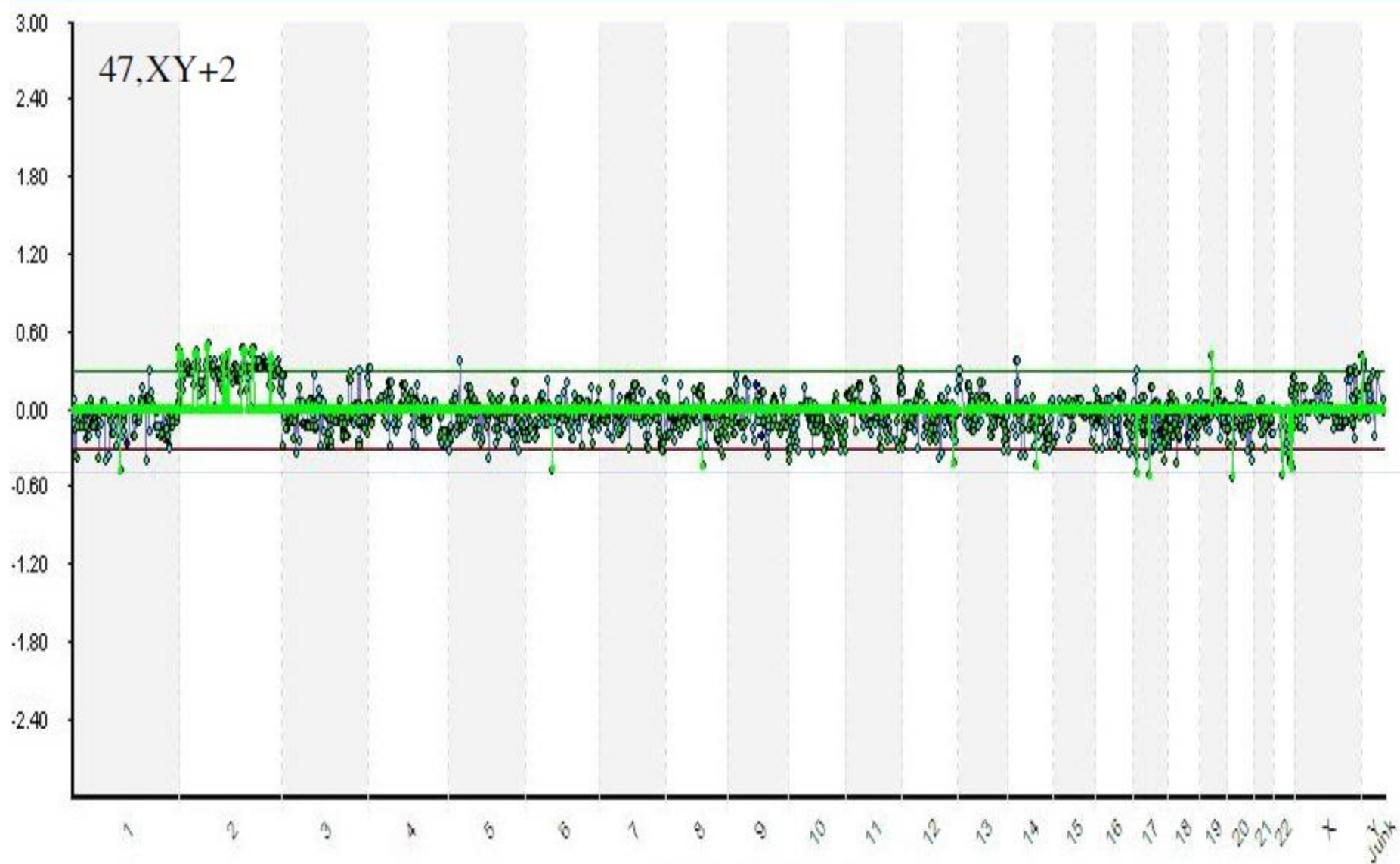
Less embryos to process

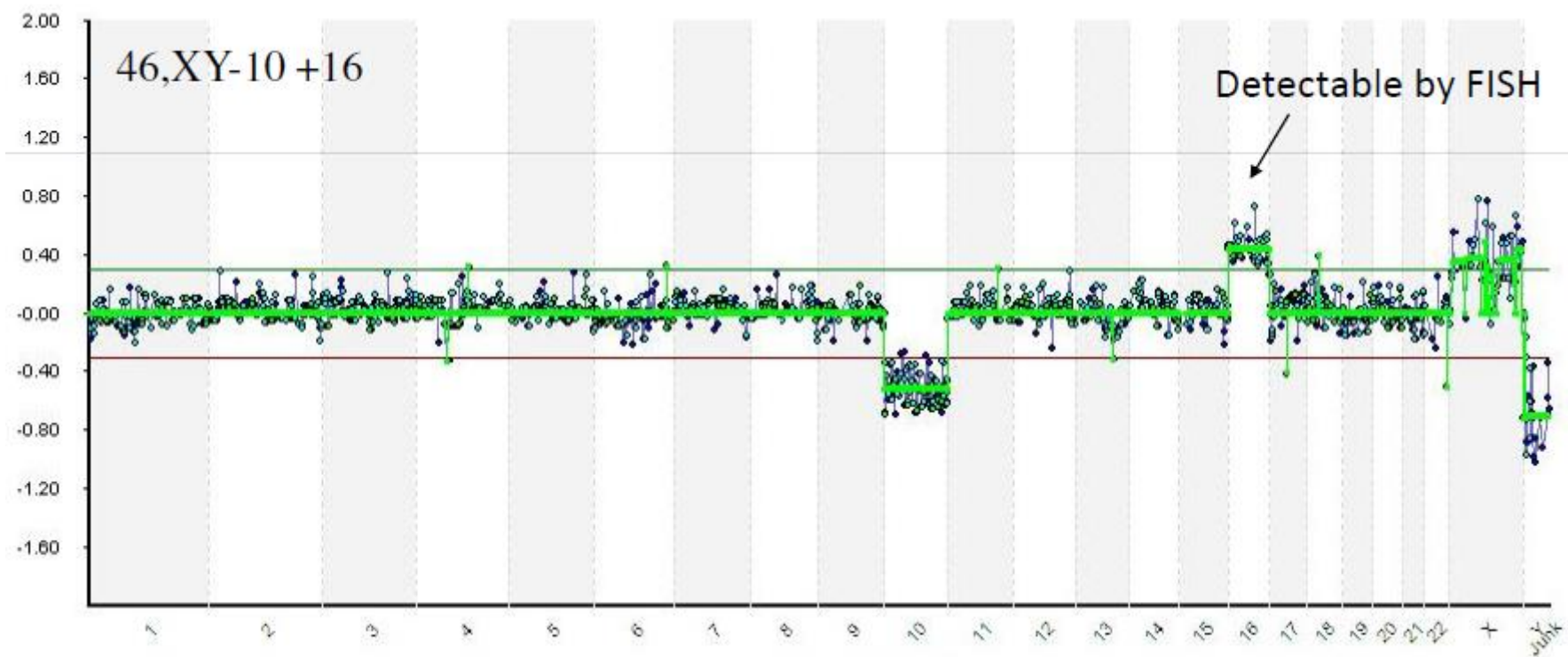
Facilitates single embryo transfer

Uterine environment optimized after thaw

24 chromosome analysis
by array technology







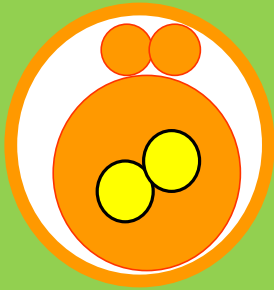
Scott

Blastocyst biopsy with comprehensive chromosome screening and fresh embryo transfer significantly **increases** in vitro fertilization implantation and delivery rates: a randomized controlled trial.

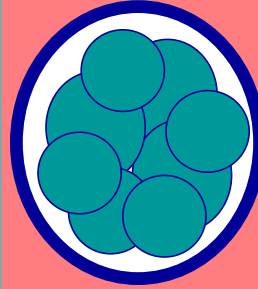
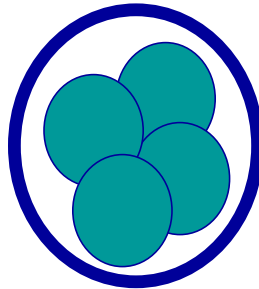
Fertil Steril. 2013

Next generation sequencing

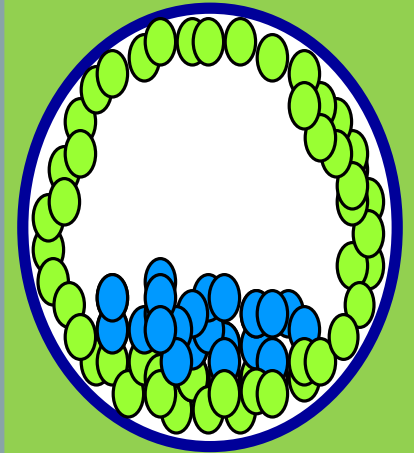
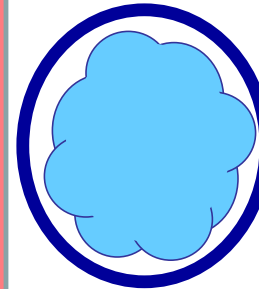
Day 1
Polarbody
Biopsy



Day 3
Blastomere
Biopsy



Day 5
Trophoblast
Biopsy



Idiopathic repeated
pregnancy loss (RPL)

Preimplantation genetic screening (PGS)

Werlin

Preimplantation genetic diagnosis (PGD) as both a therapeutic and diagnostic tool in assisted reproductive technology.

Fertil Steril 2003

Munné

Preimplantation genetic diagnosis reduces pregnancy loss in women 35 and older with a history of recurrent miscarriages.

Fertil Steril 2005

Garrisi

Effect of infertility, maternal age, and number of previous miscarriages on the outcome of preimplantation genetic diagnosis for idiopathic recurrent pregnancy loss.

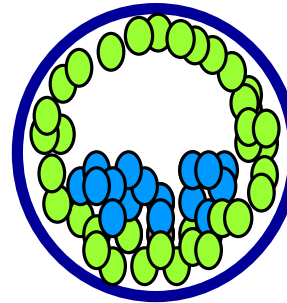
Fertil Steril 2009

Rubio

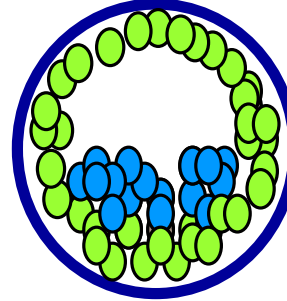
Prognosis factors for Preimplantation Genetic Screening in repeated pregnancy loss.

Reprod Biomed Online 2009

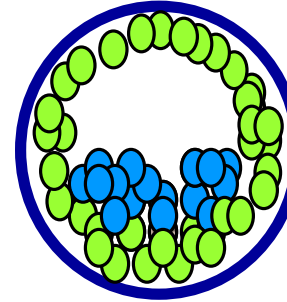
Down – Syndrome



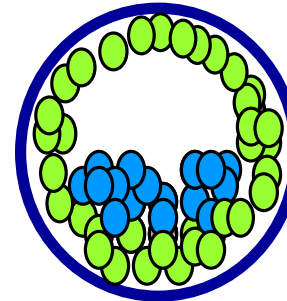
BRCA 1 Mutation
Homozygotic



Myopia



Schizophrenia



Monogenetic diseases

Preimplantation genetic diagnosis (PGD)

Affected first child

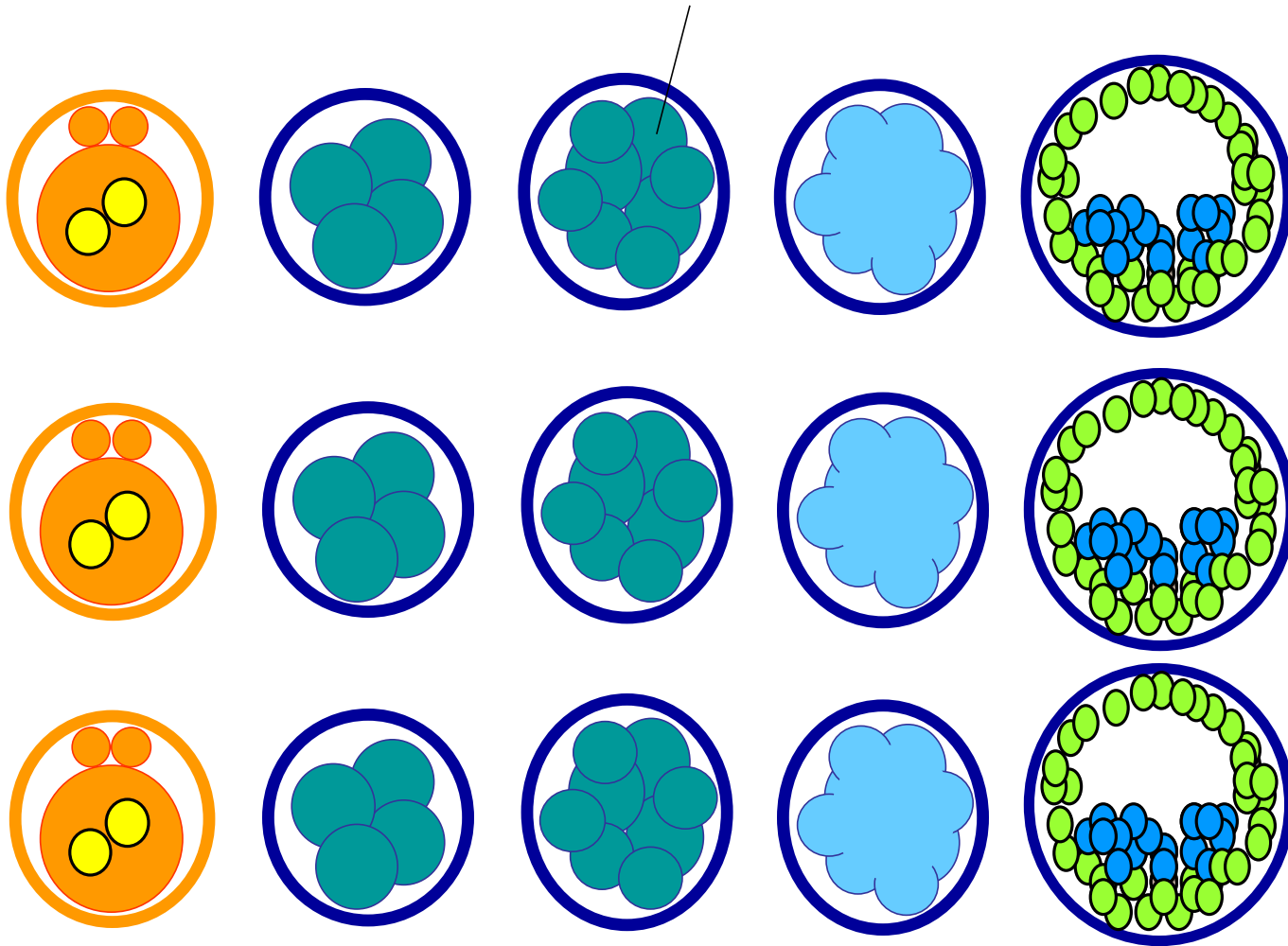
Designerbaby
„Saviour sibling“

Kahraman S

Successful haematopoietic stem cell transplantation in 44 children from healthy siblings conceived after preimplantation HLA matching.

Reprod Biomed Online. 2014

HLA – System



Beta-thalassaemia

Wiskott-Aldrich syndrome

Fanconi anaemia

Sickle cell anaemia

Acute myeloid leukaemia

Acute lymphoblastic leukaemia

Glanzmann's thrombasthenia

Diamond-Blackfan anaemia

X-linked adrenoleukodystrophy

Mucopolysaccharidosis type I

Würfel W

Pregnancy and Birth After a Two-Step
PGD: Polar Body Diagnosis for
Hemophilia A and Array CGH on
Trophectoderm Cells for Chromosomal
Aberrations.

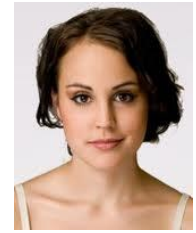
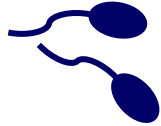
Geburtshilfe Frauenheilkd. 2013

Advanced female age

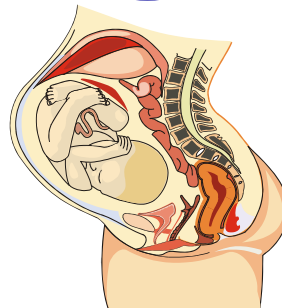
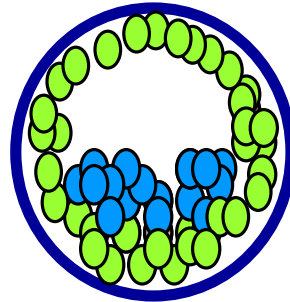
Advancing female age

Social egg freezing

2014



2037



Genetic diseases

Down syndrome

BRCA 1 Mutation

Chorea Huntington

Muscular dystrophy Duchenne

Cystic fibrosis

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Виена Ин-Витро Център
Vienna In-Vitro Center

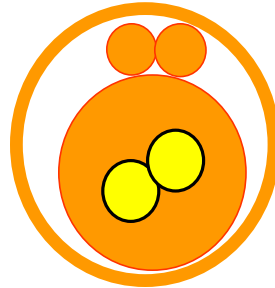
Preconception

Carrier
Screening
(Recombine)

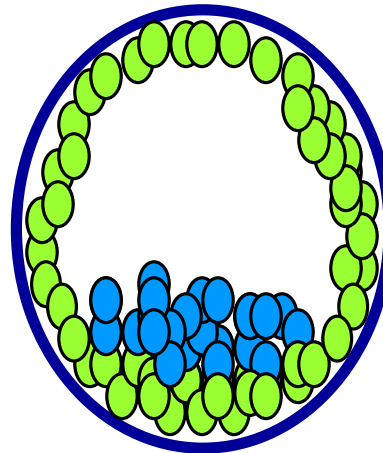


Assisted Reproduction

Day 1
Polarbody
Biopsy



Day 5
Trophoblast
Biopsy



Early pregnancy

Non Invasive
Prenatal testing
(Prena, NIPT)

