

# **Cytogenetics III:** Changes in chromosome number

Genetics 371B Lecture 17

27 Oct. 1999

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**Euploid:** normal chromosome sets

**Aneuploid:** incomplete (unbalanced) chromosome sets

◆ In humans—aneuploidy in up to 35% of spontaneous abortions (6–20 weeks)

◇ Monosomy:  $2n - 1$

Human (females) — only one kind of monosomy...

*1 in 20000 live births*

◇ Trisomy:  $2n + 1$

Most common (at conception ?)— chr 16

Most common at live birth— trisomy 21 —Down syndrome  
*1 in 750 live births*

◇ Less common:

trisomy 18 (1 in 10000)

trisomy 13 (1 in 20000)

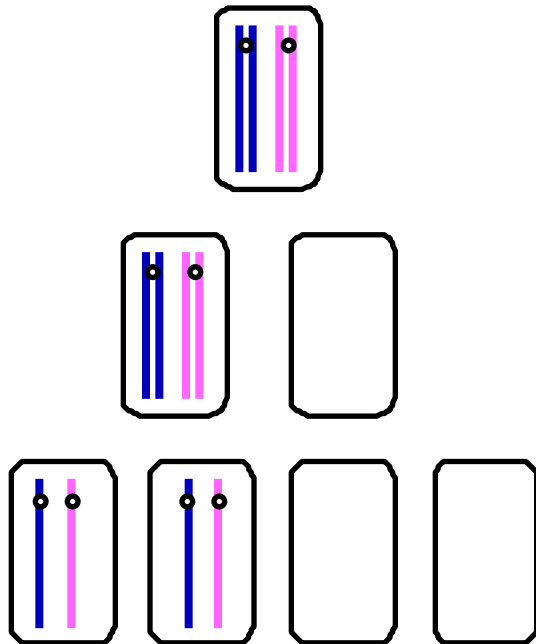
◇ Why better survival with trisomy 21 than other trisomies?

## **Hierarchy of tolerance** of aneuploidy

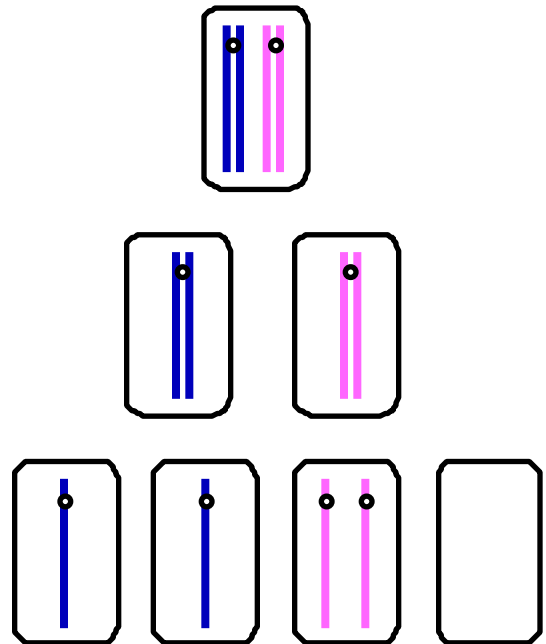
- ◇ sex chromosome aneuploidy > autosomal aneuploidy;
- ◇ autosomal triploidy > monosomy

Major cause of aneuploidy:  
**nondisjunction** during meiosis

...can occur at Meiosis I



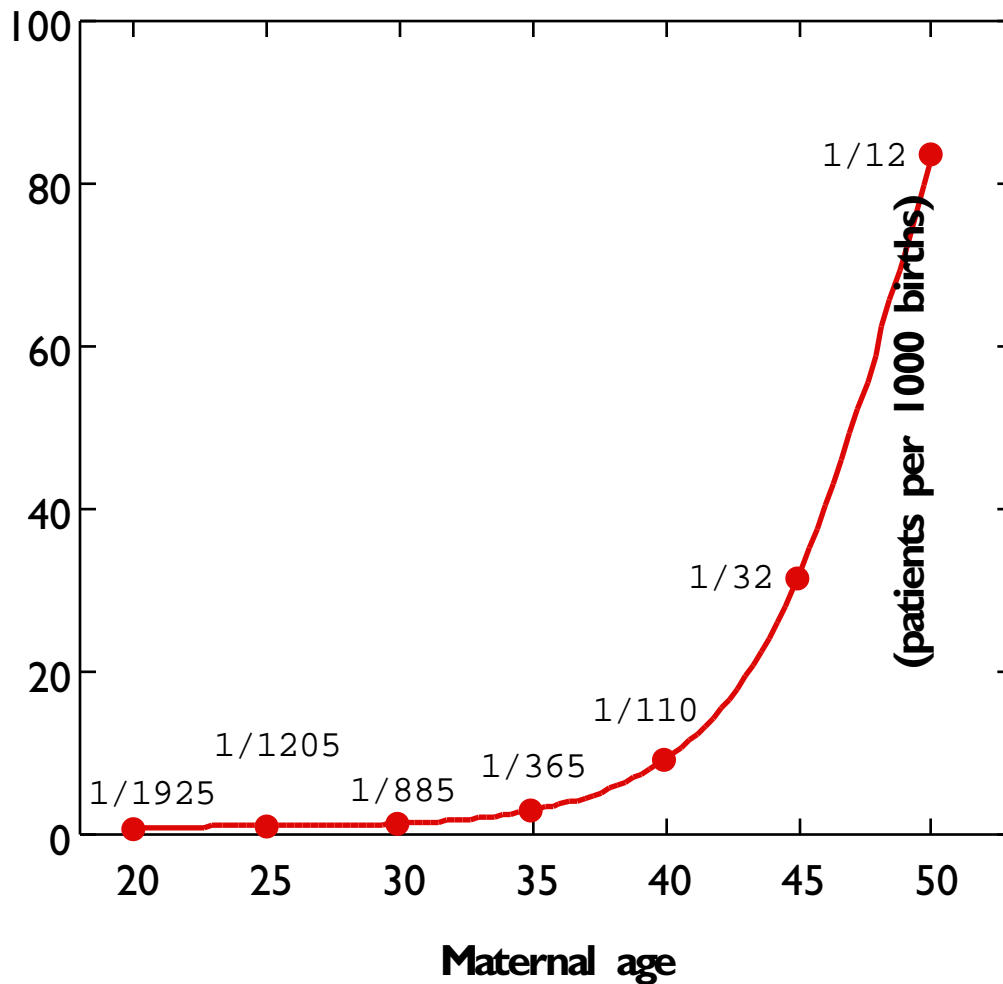
... or at Meiosis II



### **Consequences:**

- ◆ Defective products
  
- ◆ Allele composition

# Aneuploidy and maternal age



Estimated Down syndrome frequency

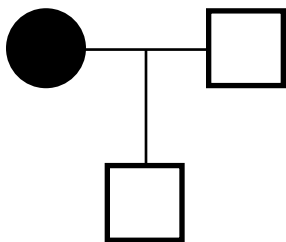
## Why?

- ◆ ND ↑ in older oocytes? Checkpoints?
- ◆ less robust spindle?
- ◆ increasing pool of “poor” oocytes?

About 20–25% of Down syndrome cases –  
**paternal** nondisjunction

Aneuploidy from **maternal** or **paternal**  
nondisjunction? Sometimes, clues from the pedigree...

X<sup>g</sup> = X-linked recessive condition  
Paternal or maternal ND here?

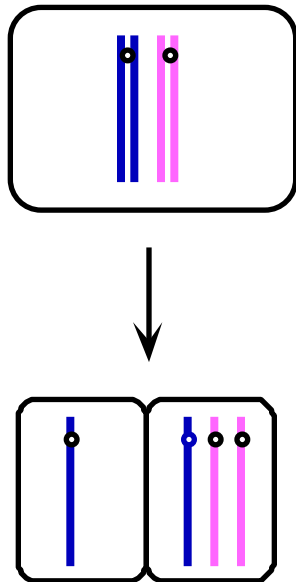


Klinefelter (XXY) male

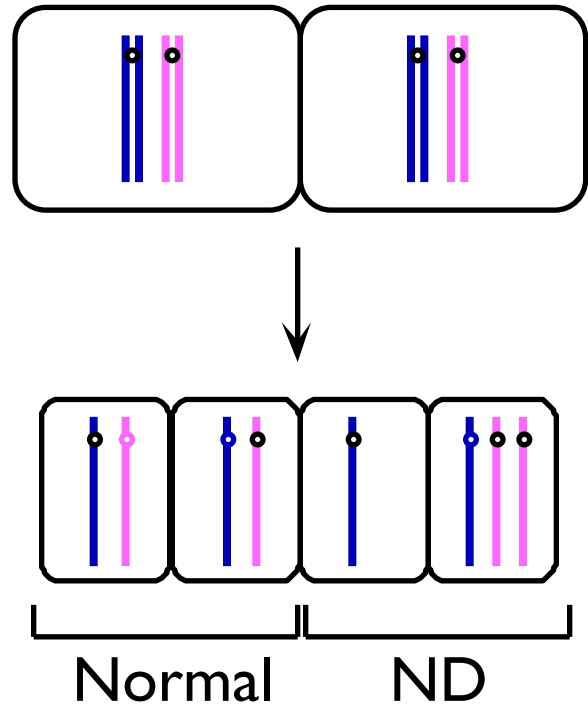
# Mitotic nondisjunction

e.g., Down syndrome mosaics

ND in 1st cleavage



ND after 1st cleavage



# Ploidy changes

- ◆ **Plants:** It's not all bad news... polyploidy is often desirable
  - ◇ Polyploids larger
  - ◇ Infertility due to polyploidy
  
- ◆ **Animals:** Haploids, polyploids rare

Triploidy in humans –