



Genetics = the study of *heredity*

***Genes* = the “matter” of heredity**



Aristotle (384-322 B.C.):

Inheritance = physical substance coming from both parents

Giraffe = hybrid animal, resulting from cross between leopard & camel





Honore Daumier: “Un Nouveau Nez”



Pen & Ink Sketches No. 7



London J. L. Marks

A CHIP OF THE OLD BLOCK.

*My gracious! here's a likeness,— why it's
Daddy all over obeys its little soul.*



“The fittest family”



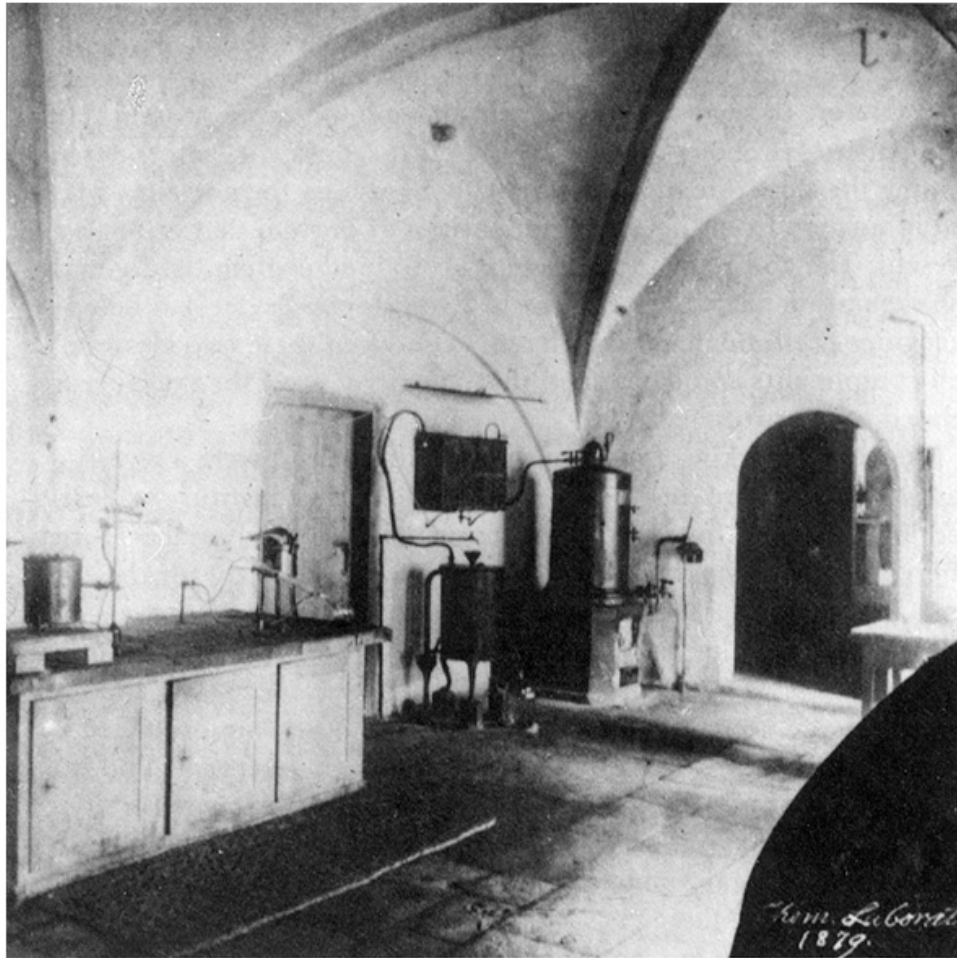
Eugenics Record Office: “breeding” better humans



Experiments in Plant Hybridization 1865

“differentiating characters”

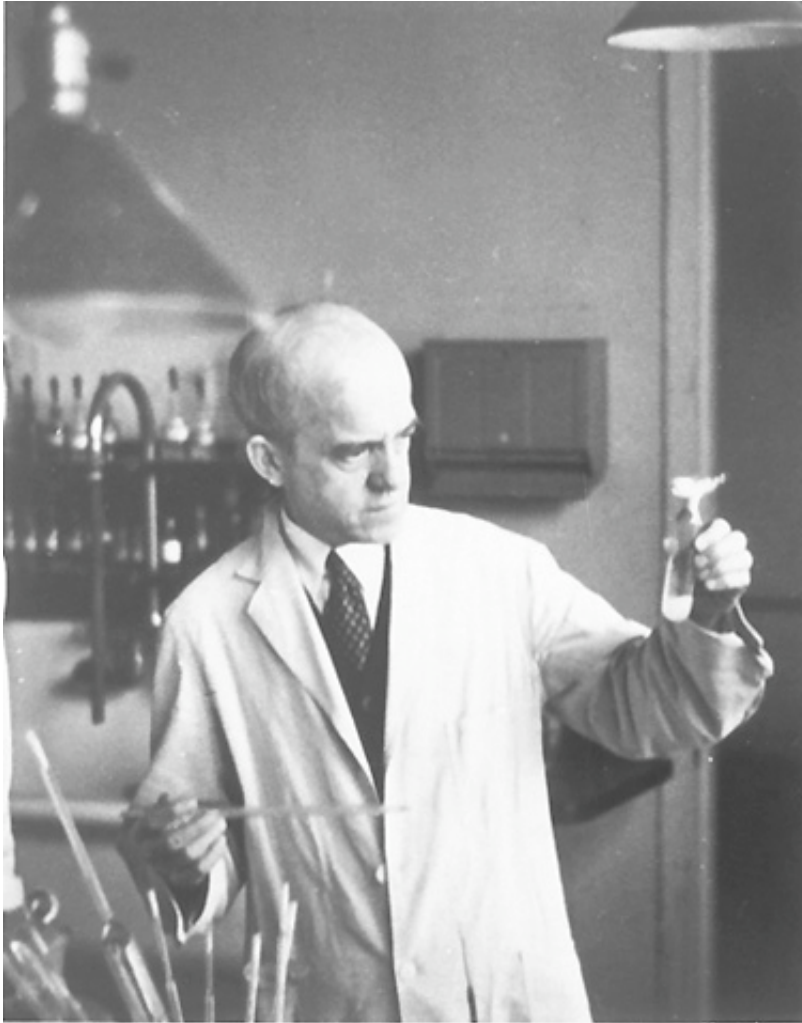




Friedrich Miescher

**1869:
nuclein (DNA) from pus**





**Oswald Avery, Colin Macleod,
Maclyn McCarty**

1940's

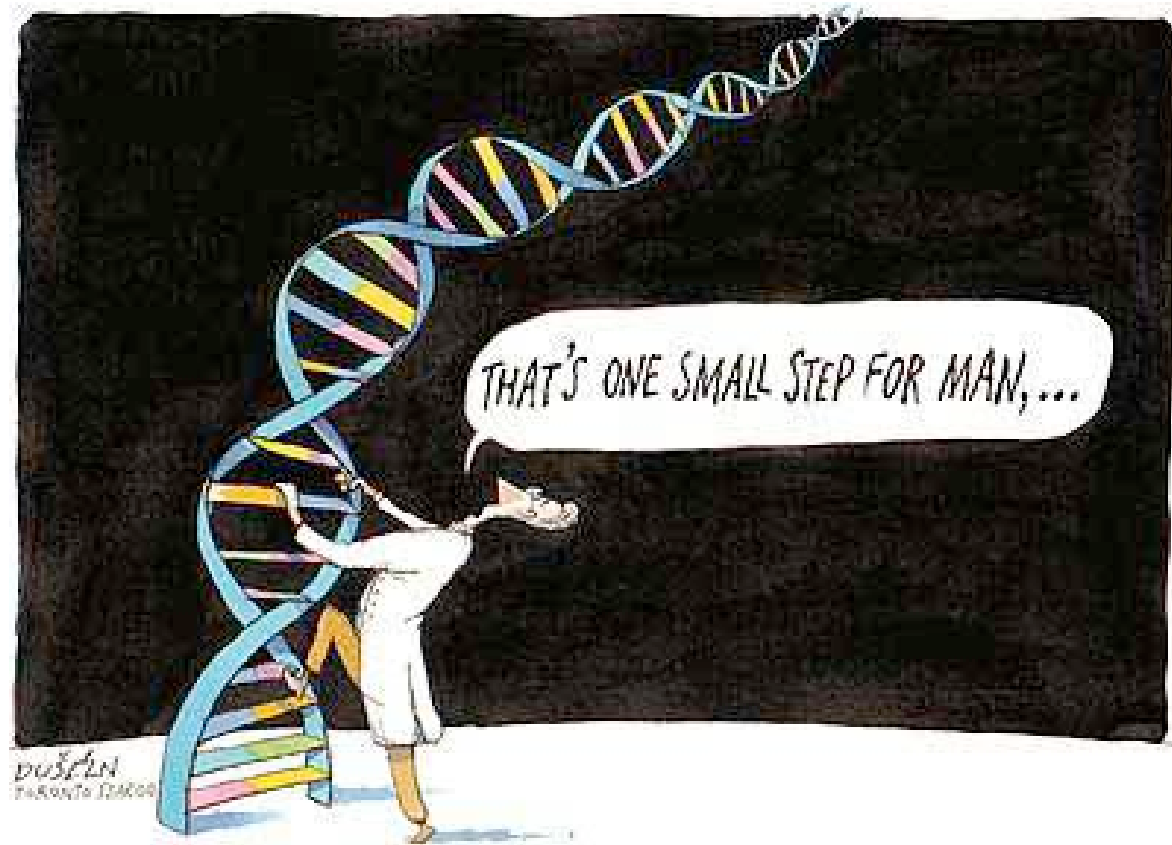
gene = DNA

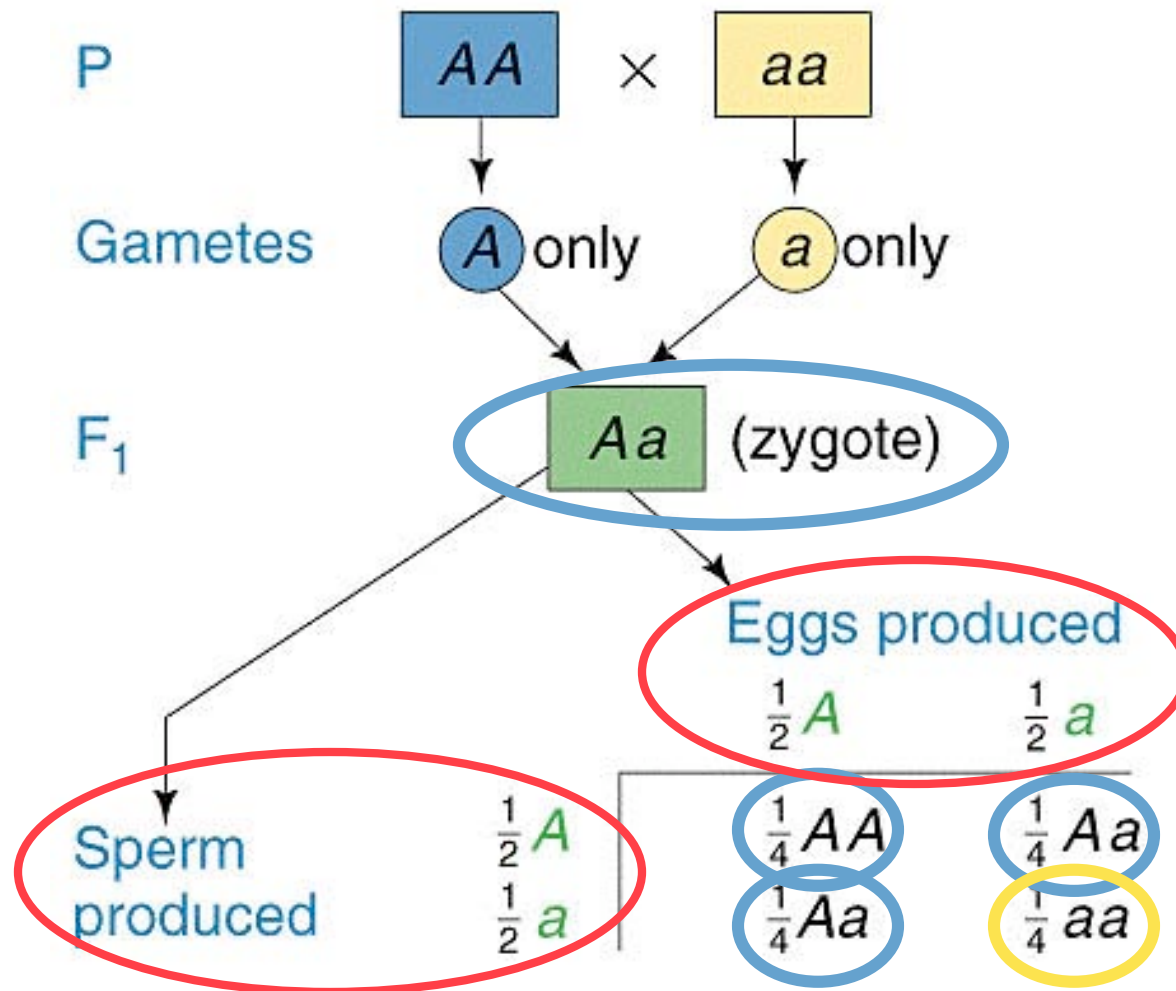




Watson & Crick, 1953



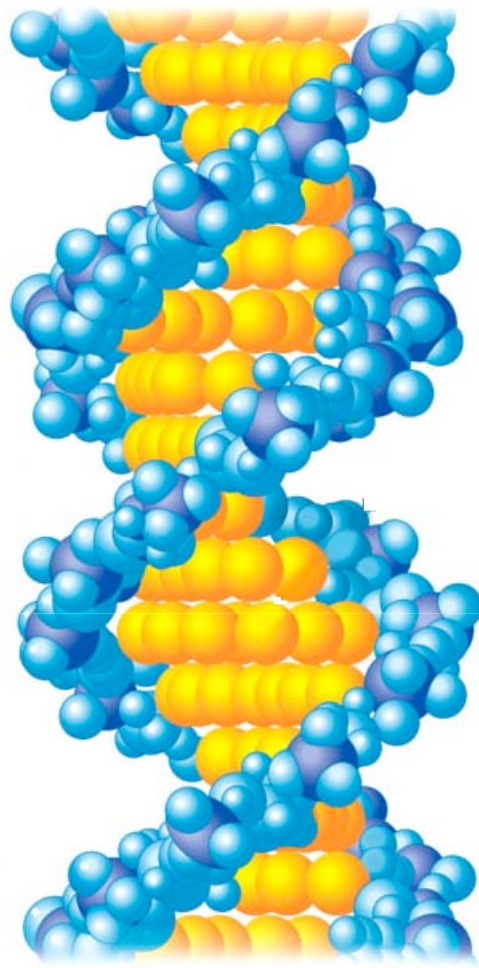


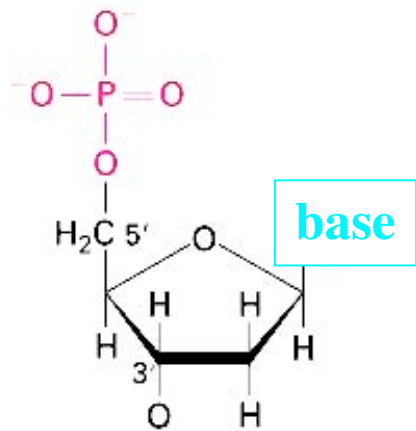


Genes: Overall F₂ ratio

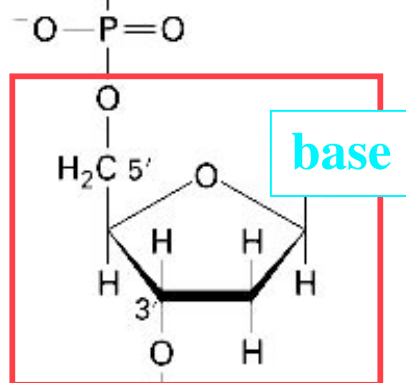
Traits: Overall F₂ ratio





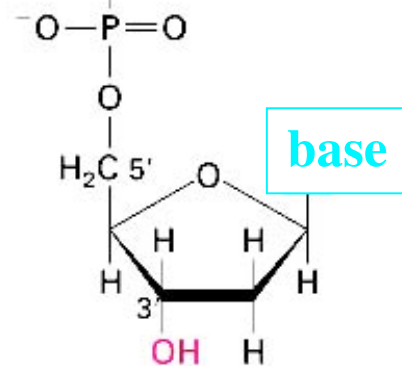


DNA sugar-phosphate backbone



deoxyribose

(deoxyribonucleic acid)

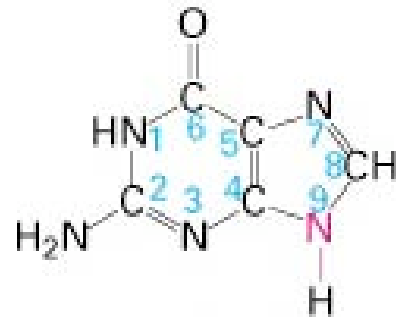


DNA bases

PURINES



Adenine (A)



Guanine (G)

PYRIMIDINES

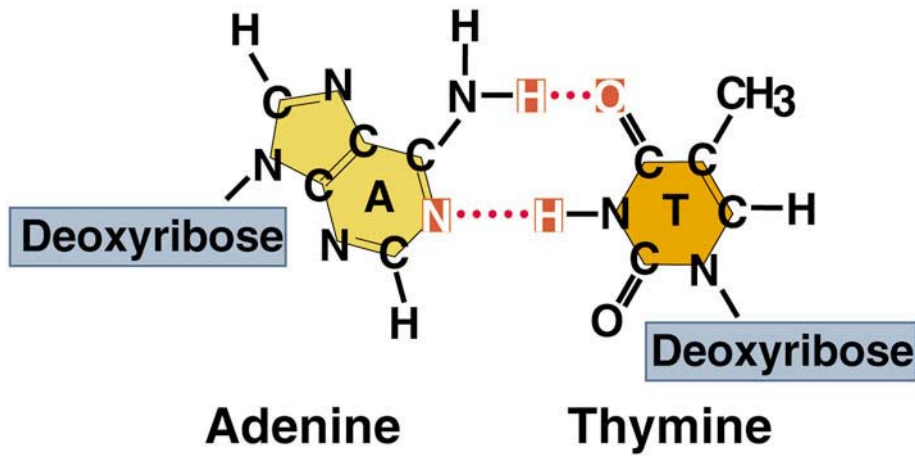


Thymine (T)

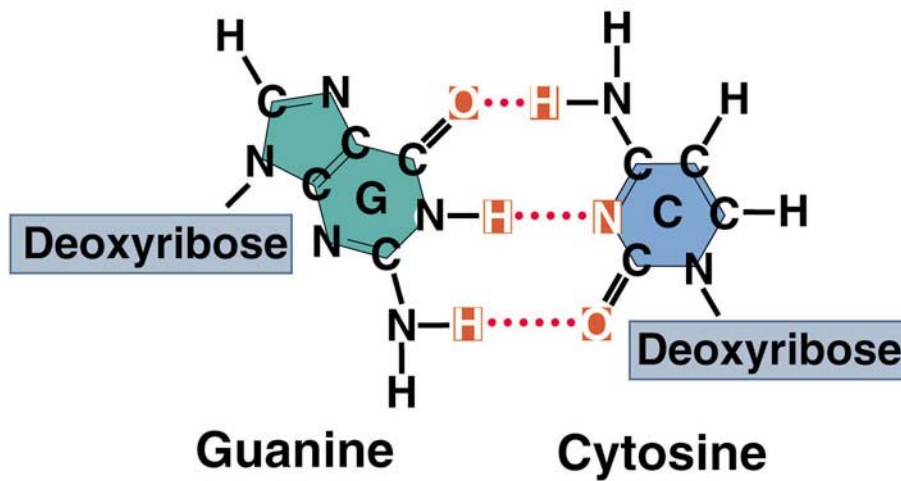


Cytosine (C)





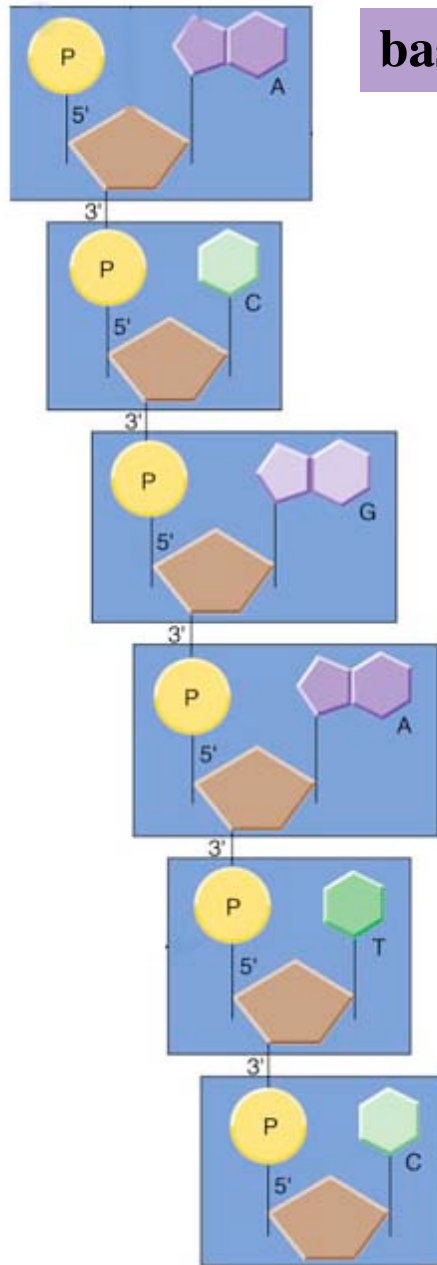
DNA base pairing



phosphate

sugar

base

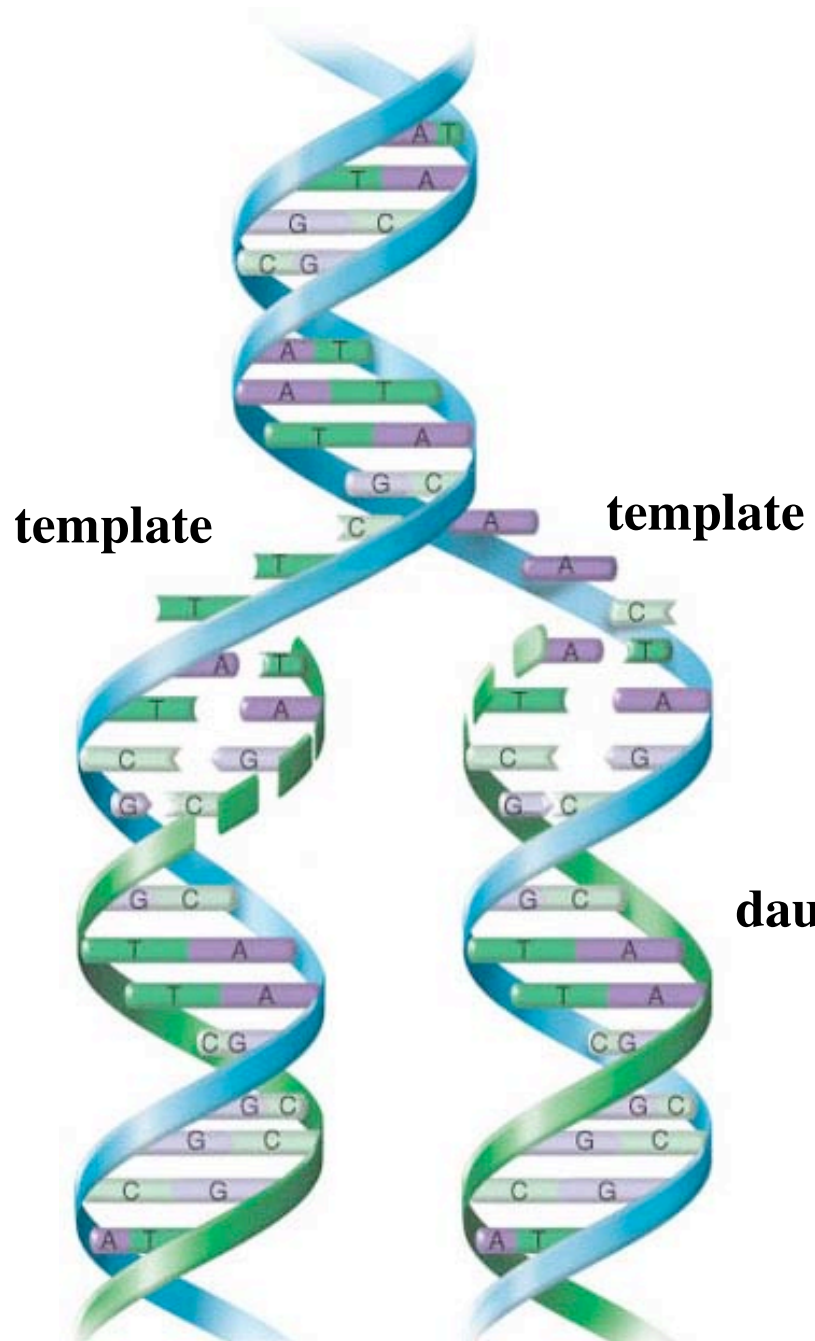


DNA polymer

(nucleotide polymer)



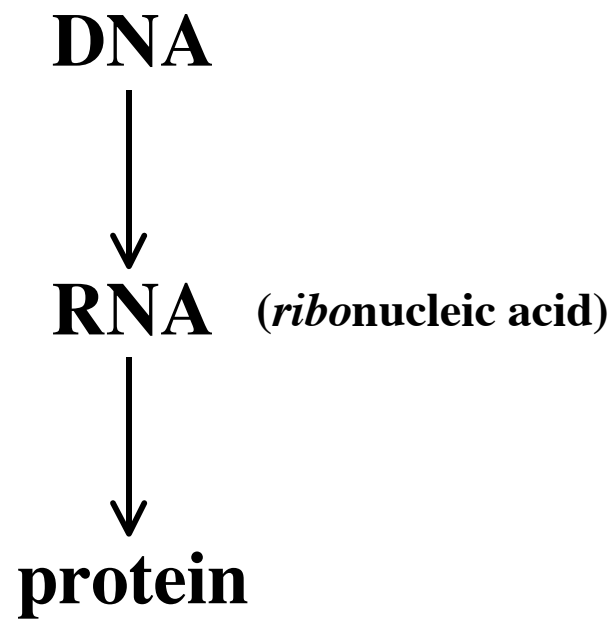




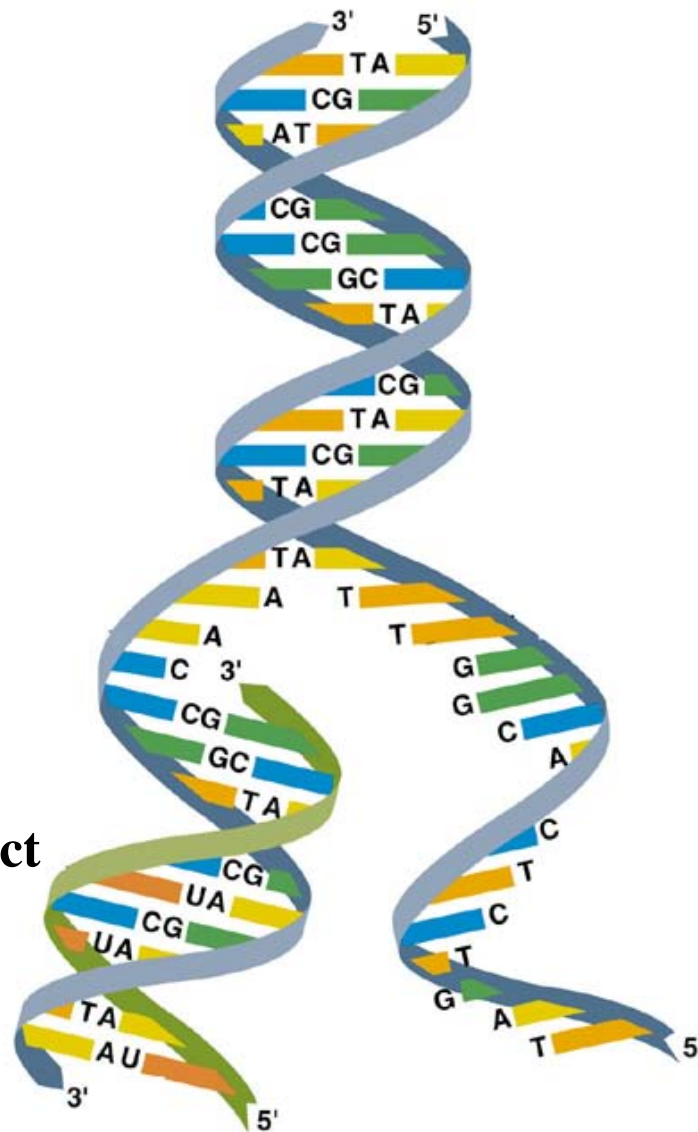
replicating DNA



The Central Dogma

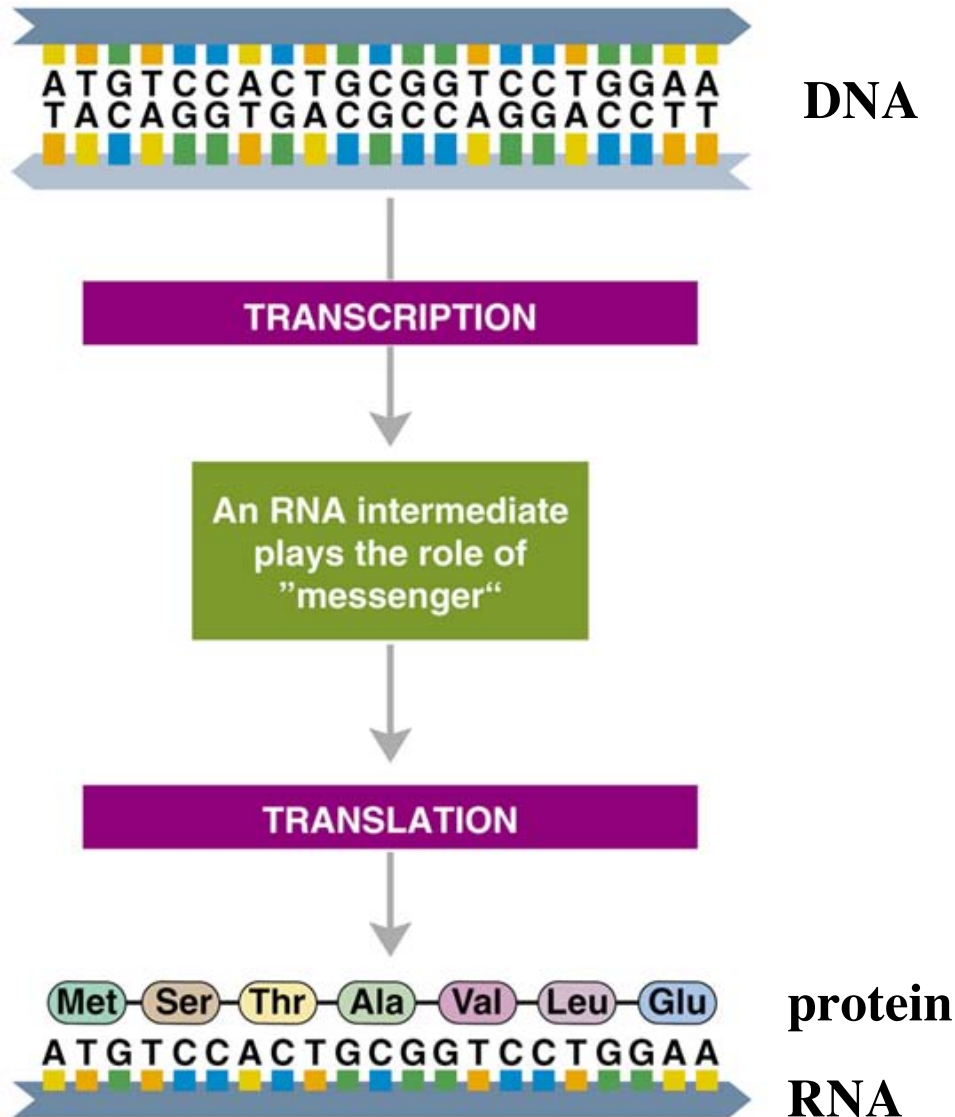


RNA product



DNA being transcribed

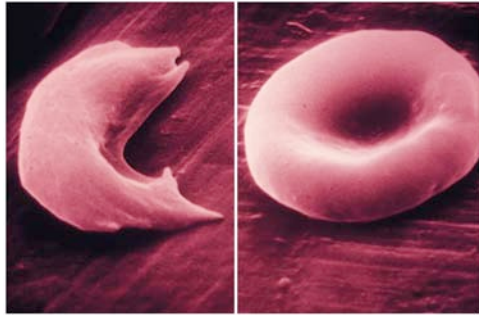




		Second letter					
		U	C	A	G		
First letter	U	UUU } Phe UUC } UUA } Leu UUG }	UCU } UCC } Ser UCA } UCG }	UAU } Tyr UAC } UAA Stop UAG Stop	UGU } Cys UGC } UGA Stop UGG Trp	U C A G	Third letter
	C	CUU } CUC } Leu CUA } CUG }	CCU } CCC } Pro CCA } CCG }	CAU } His CAC } CAA } Gln CAG }	CGU } CGC } Arg CGA } CGG }	U C A G	
	A	AUU } AUC } Ile AUA } AUG Met	ACU } ACC } Thr ACA } ACG }	AAU } Asn AAC } AAA } Lys AAG }	AGU } Ser AGC } AGA } Arg AGG }	U C A G	
	G	GUU } GUC } Val GUA } GUG }	GCU } GCC } Ala GCA } GCG }	GAU } Asp GAC } GAA } Glu GAG }	GGU } GGC } Gly GGA } GGG }	U C A G	

the Genetic Code





sickle cell
rbc

normal
rbc

defective gene: mutant hemoglobin

normal DNA GTG CAC CTG ACT CCT **GAG** GAG AAG TCT

sickle cell DNA GTG CAC CTG ACT CCT **GTG** GAG AAG TCT

normal Hb VAL HIS LEU THR PRO **GLU** GLU LYS SER

sickle cell Hb VAL HIS LEU THR PRO **VAL** GLU LYS SER



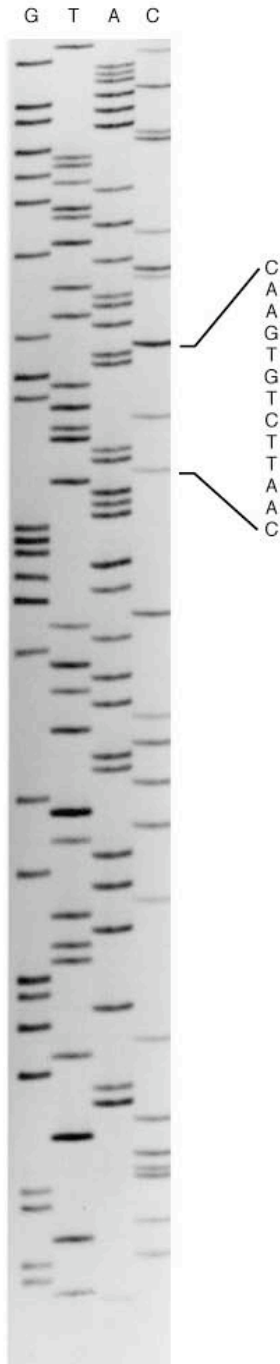
genetic testing (screening) =

the systematic search for individuals who have certain genetic constitutions (genotypes) that (1) are already associated with disease or predisposition to disease or (2) may lead to disease in their descendants

e.g.:

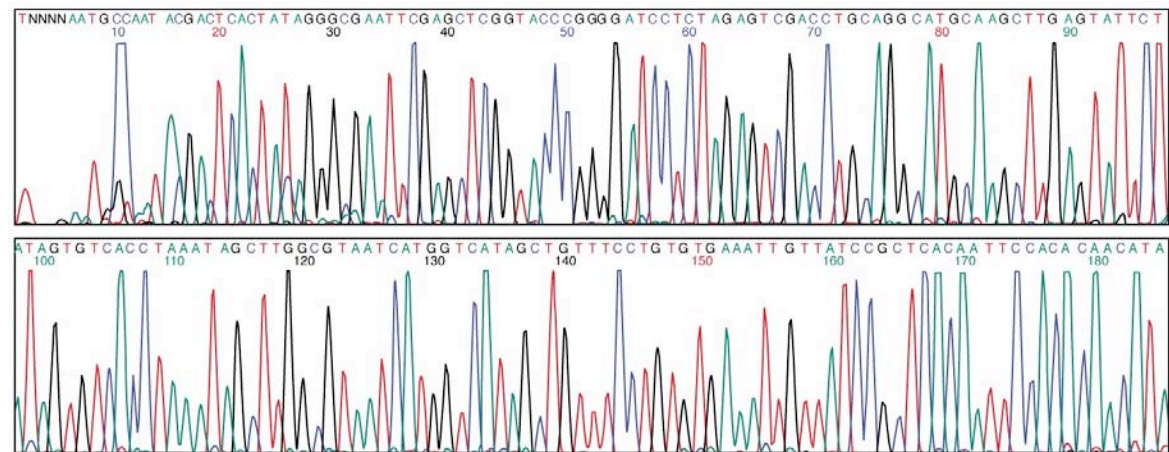
- **newborn screening for inherited metabolic diseases**
- **carrier detection for diseases like Tay-Sachs**





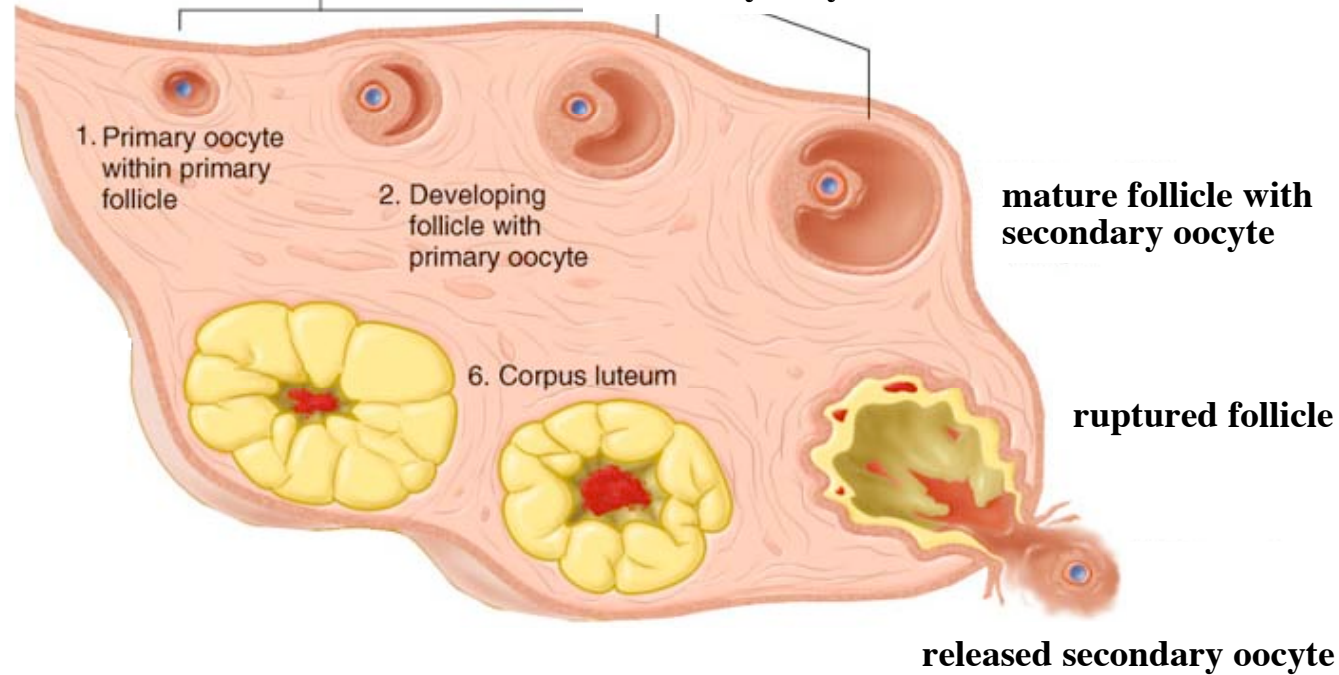
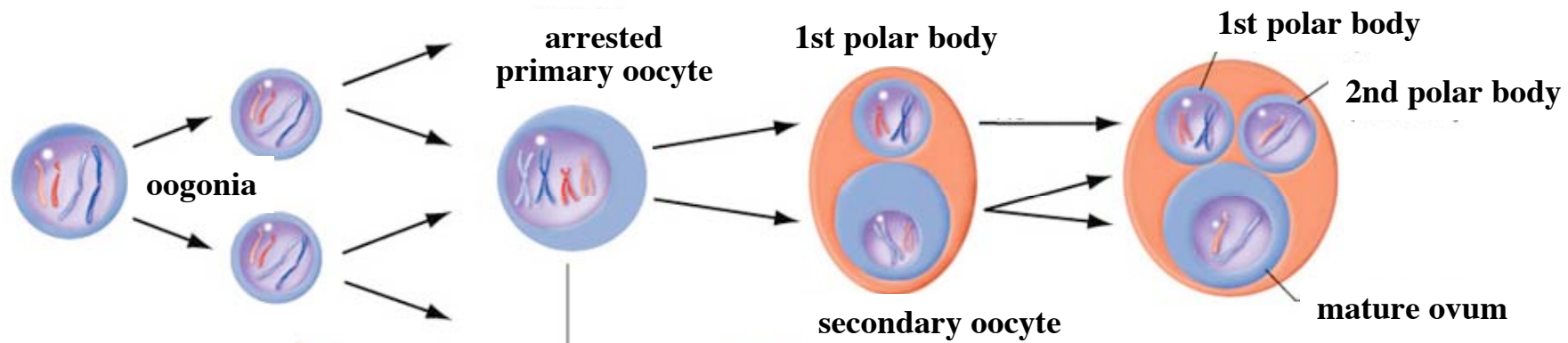
genetic tests

- functional/structural assays of proteins
[phenylketonuria: phenylalanine hydroxylase]
- direct analysis of DNA

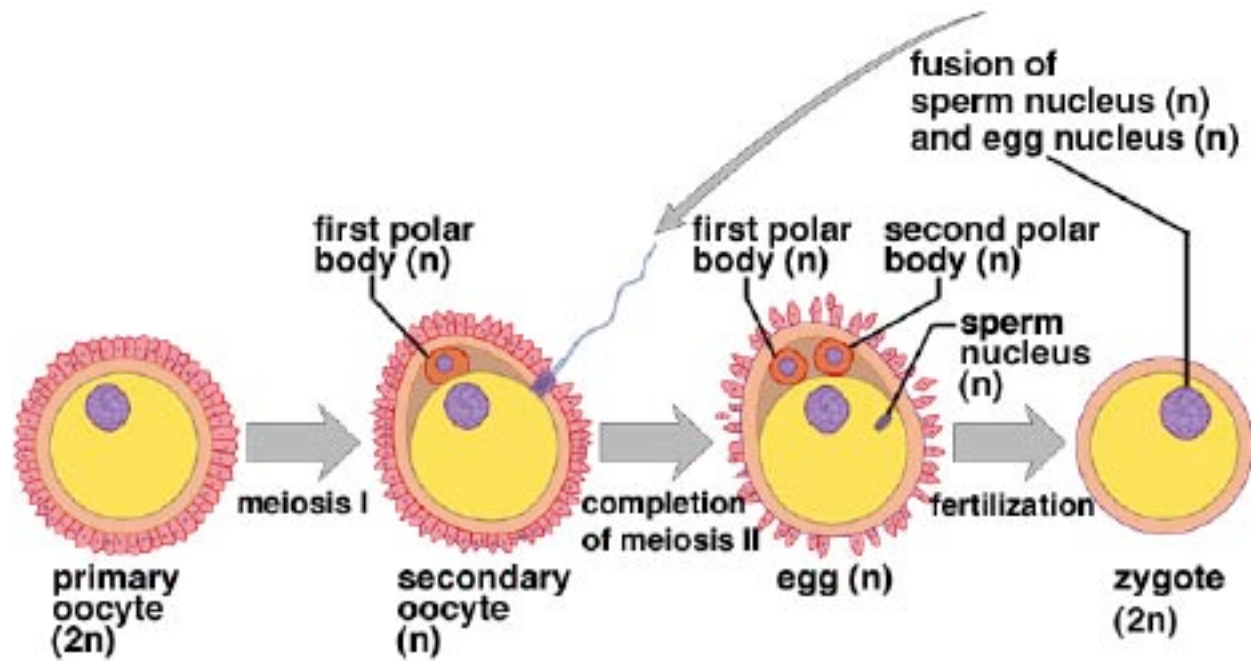


oogenesis

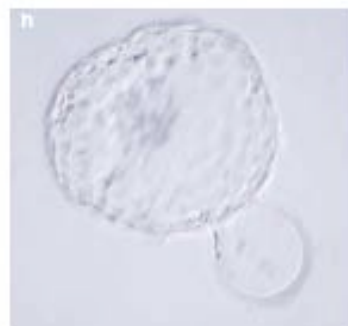
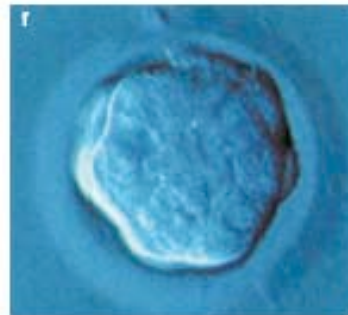
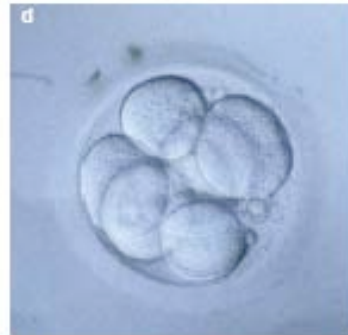
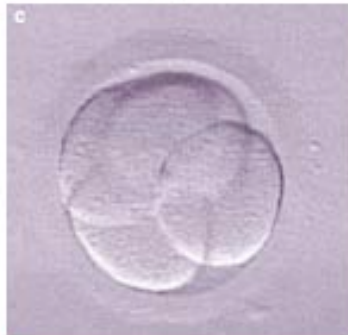
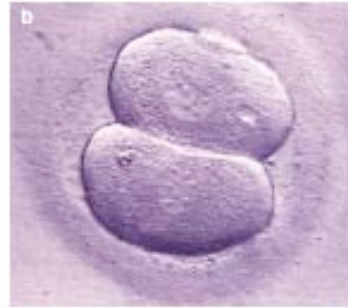
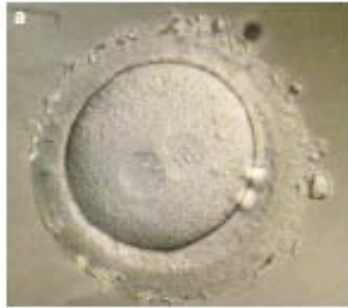
(meiosis)



oogenesis & fertilization



zygote



preimplantation development

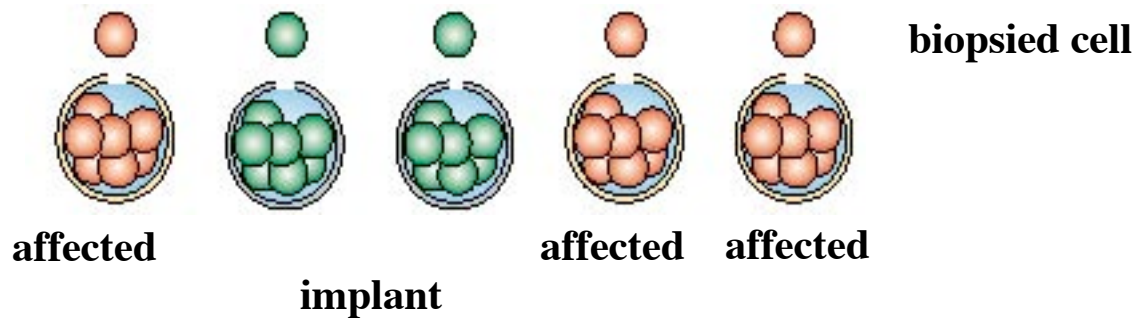
0 through 6 days post fertilization



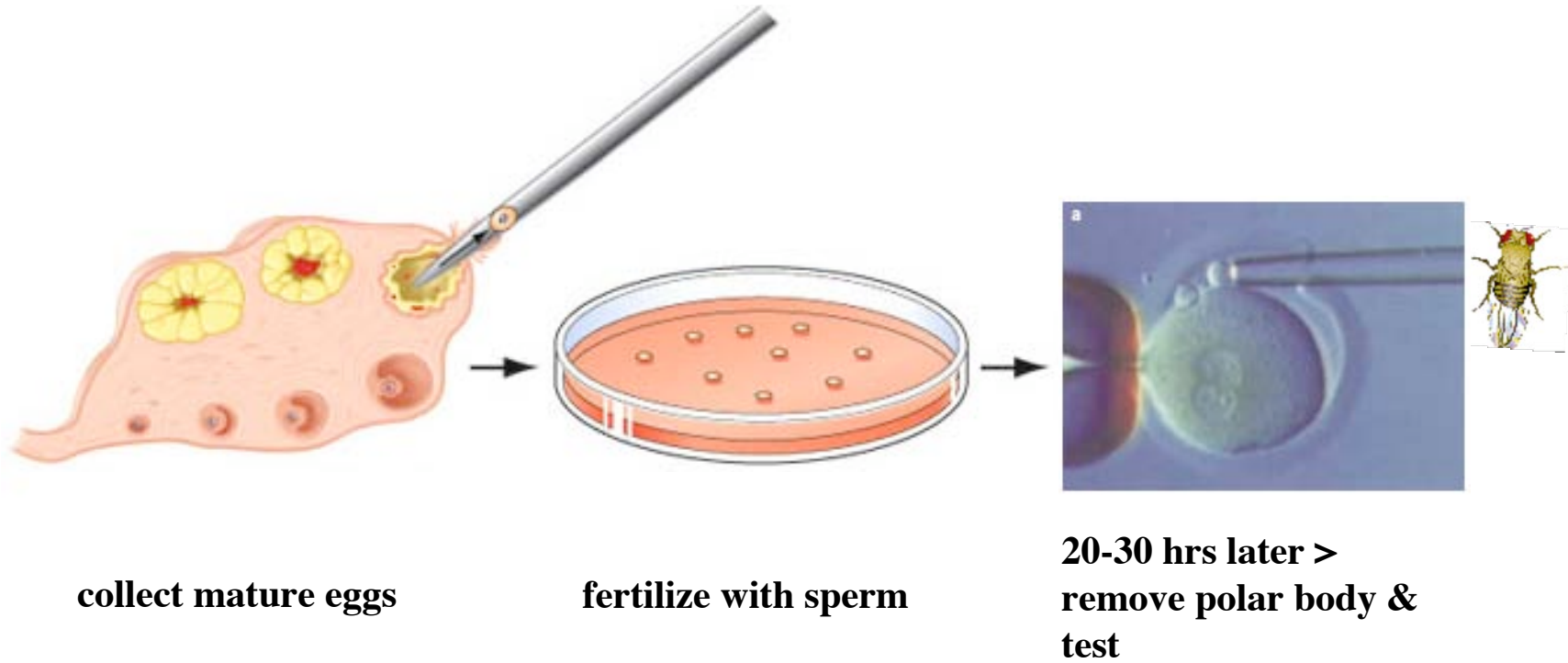
preimplantation genetic diagnosis (PGD)

in vitro fertilization (IVF) followed by biopsy

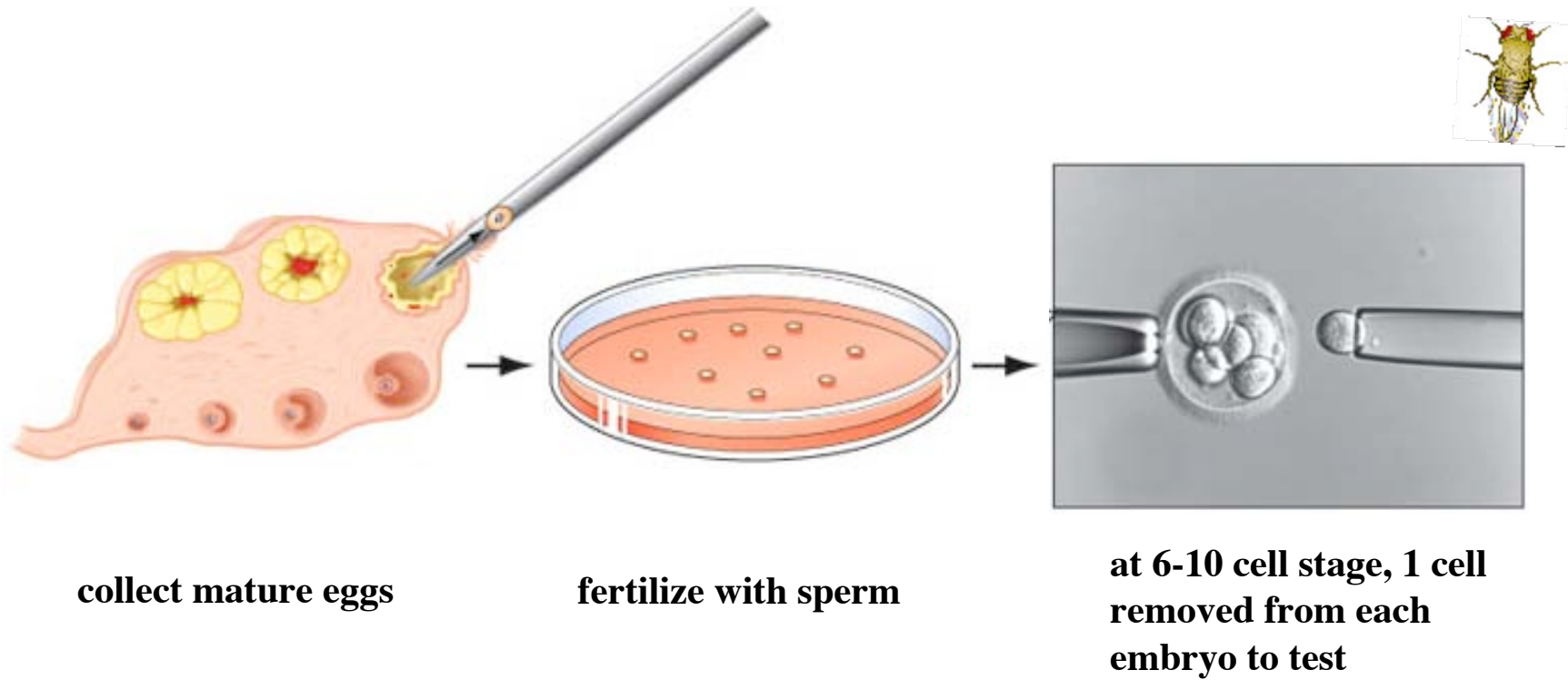
- **polar body biopsy**
 - **infer embryo's genetic condition**
 - **cannot learn about paternal contribution**
- **cleavage stage biopsy**
 - **test embryonic cells directly**



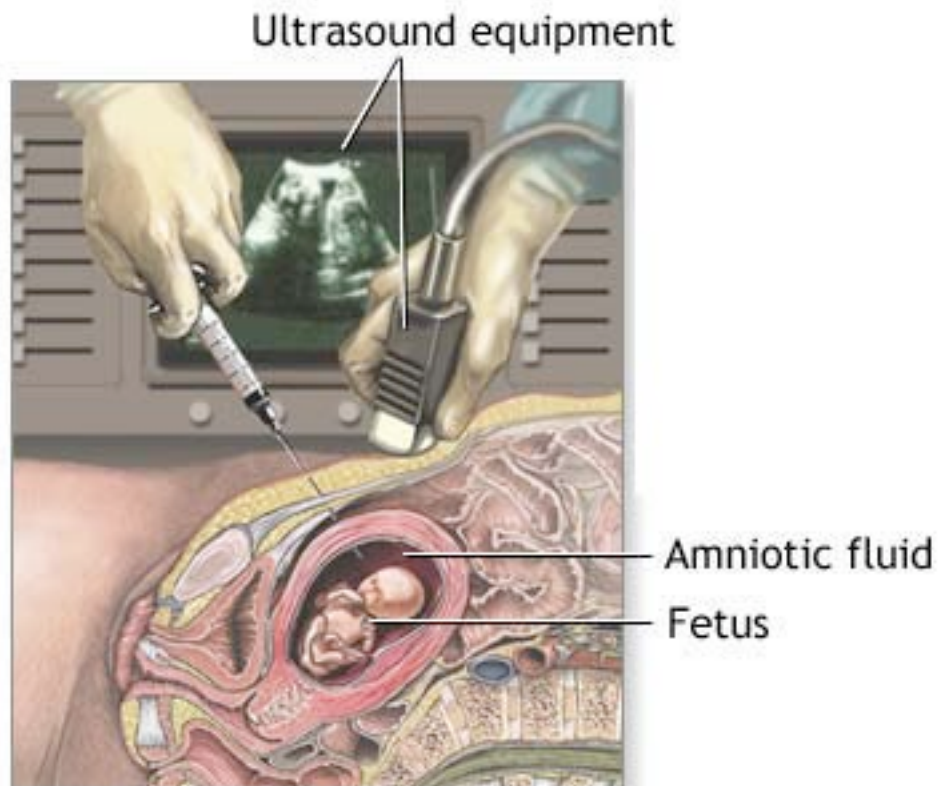
polar body diagnosis



cleavage stage diagnosis

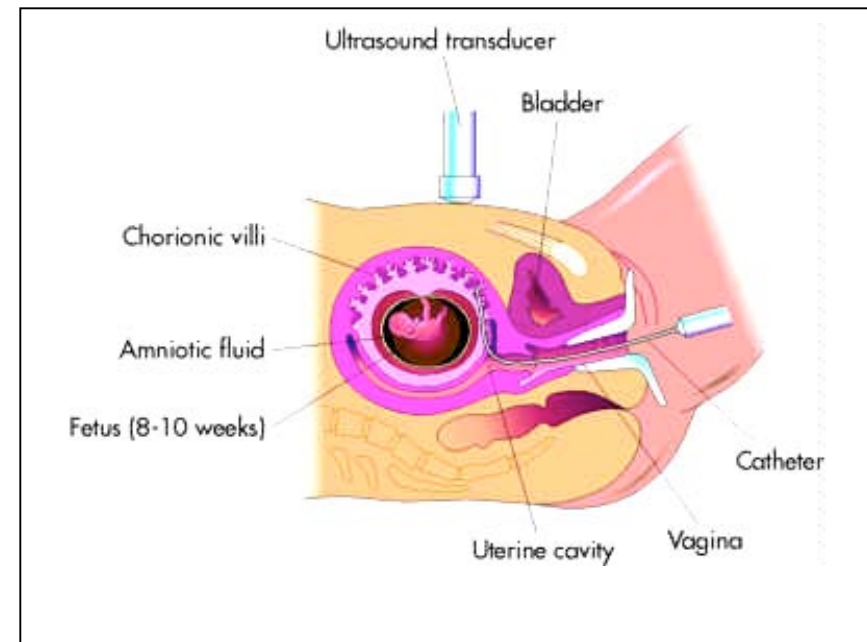


postimplantation genetic diagnosis



amniocentesis

chorionic villus sampling (CVS)





Where do we go from here?

How will we use the information we can gather about our genetic material?