Genetics 5

- Answer to iClicker 5A
- Pedigrees (symbols)
- Inherited genetic disease
  - autosomal recessive (PKU)
  - autosomal dominant (Marfan syndrome)
  - sex-linked recessive (color-blindness)
- Answer to iClicker 5B
- Due in Lab this week
  - Pre-Lab 2
  - VGL 1 lab report
  - Bring textbook to lab

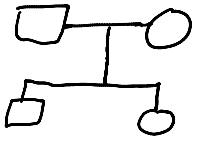
Pedigrees → way to look many crosses when have few offspring

Symbols: show phenotype → identify model & genotype

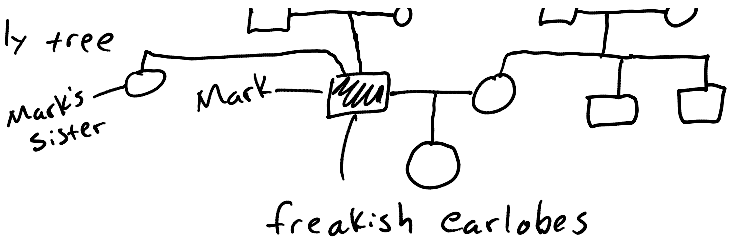
		males	females
unaffected	"normal"		
affected	"disease"		

parents

siblings



Mark's family tree



Inherited diseases

ex. phenylketonuria (PKU)
inherited intolerance to phenylalanine

controlled by 1 gene on autosome #12 ; 2 alleles

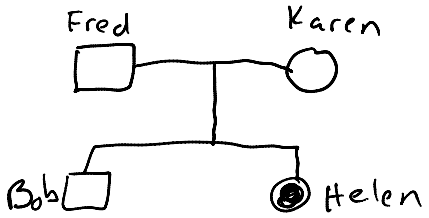
allele	contr. to phenotype
D	unaffected (dom.) } simple dominance
d	PKU (rec.) } simple dominance

because PKU is on an autosome -> autosmal recessive disease

ex. Fred (unaffected) X Karen (unaffected)

unaffected son & PKU daughter

pedigree



genotypes ① Helen is dd

② Fred & Karen must be Dd -> carriers

③ Bob DD or Dd -> (D-)

Features of Autosomal Recessive Diseases

- usually rare
- have carrier -> unaffected heterozygote (Dd)
- unaffected parents can have affected children
- if both parents affected ALL children affected

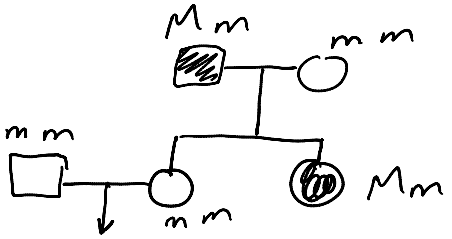
Autosomal Dominant Disease -> Marfan syndr

Autosomal Dominant Disease → Marfan Syndrome

- tall, long arms
- weak aorta → typically fatal

controlled by 1 gene on autosome #15 ; 2 alleles

allele	contr. to phenotype	
M	marfan syn. (dom.)	simple dominance
m	unaffected (rec.)	



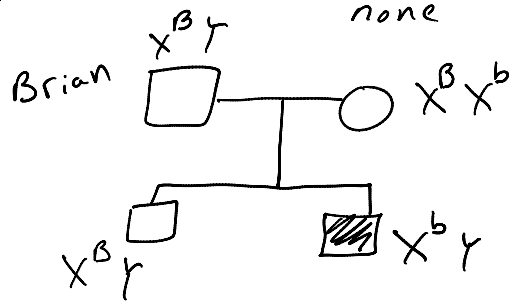
Features of Autosomal Dominant Diseases

- affected children have at least 1 affected parent
- no carriers
- if both parents are affected some children can be affected

Sex-linked reccesive → color-blindness (red-green)

controlled by 1 gene on X chromosome

allele	contr. to phenotype	
X^B	normal (dom)	simple dominance
X^b	color blind (rec.)	
Y	none	



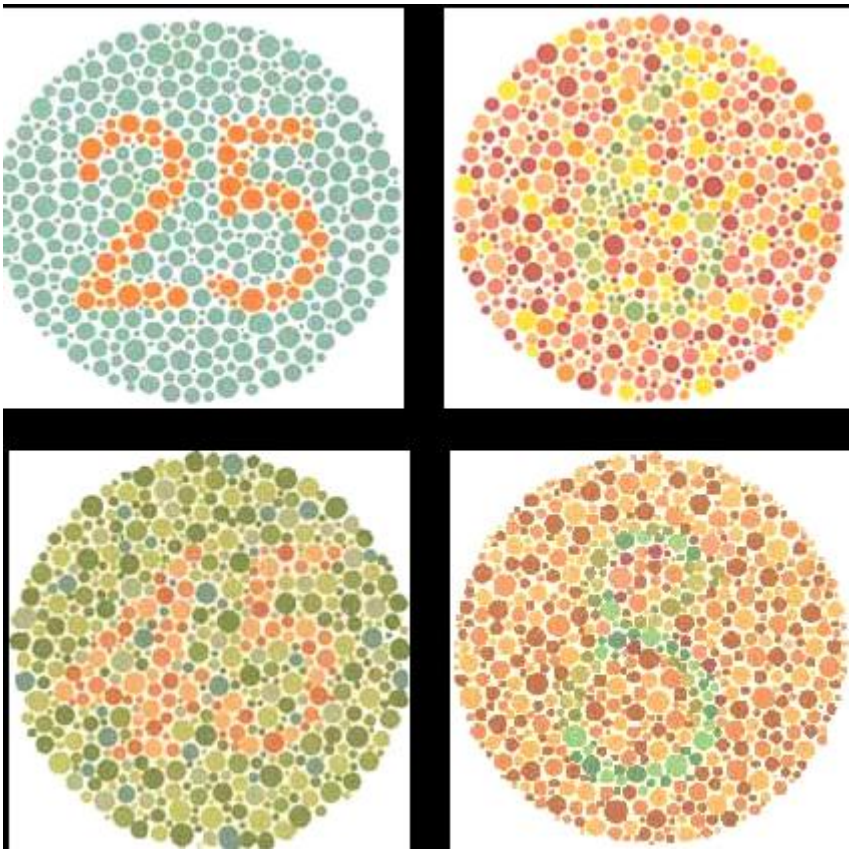
Features of Sex-linked Diseases

- usually more frequent in males
- carriers -> only female carriers
- affected mom -> all sons affected
- affected daughter -> must affected father

Boy with untreated PKU

Because a child with PKU lacks the normally functioning enzyme necessary to break down phenylalanine (PHE), it accumulates in the blood and body tissues.

This excess PHE can prevent normal brain development and result in mental retardation.



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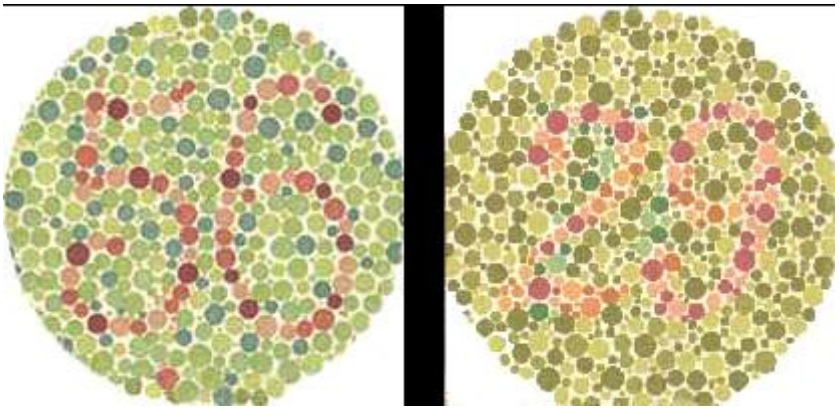

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