

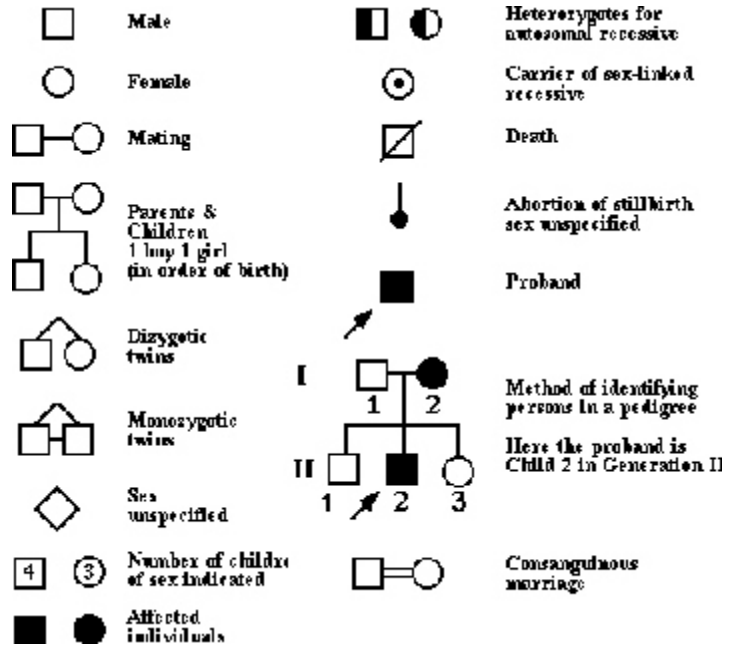
PEDIGREE CONSTRUCTION AND HUMAN TRAITS

7 Jan 1996, 10 Jan 01, 11 Jan 02, 12 Jan 04, 10 Jan 05, 9Jan09,
Griffiths *et al* 9th: 66-77 (Tab 2-28, p 41)
Klug&Cummings, 4th: p 69, Tamarin 4th p 82-87

Pedigrees shows inheritance mode, even with small #progeny.
Family tree indicates phenotype of each member's relationship
Propositus first individual to present trait

CONVENTIONS: (p 66)

female circles
male squares
unknown diamond
parents connected by horizontal line
siblings vertical line connects to parental bond, horizontal to siblings
dizygotic diag. lines connect directly to sibship line
monozygotic connected by diagonal lines to parental, single horizontal to each other
affected shaded square or circle
heterozygotes (have dominant phenotype) half shaded
arrow indicates propositus (proposita)
first individual which drew attention
roman numerals Generations are indicated by I, II, III, etc
arabic numerals number individuals within generation, arranged chronologically: 1, 2, 3, etc.



consanguineous marriage is one between relatives (incestuous if against law)

INDICATORS OF TYPES OF INHERITANCE (* MARKS BEST INDICATOR):

- Autosomal Recessive:** (p 67)
- * **if both parents affected, all children affected**
 - * often skips generations (if relatively rare)
 - * Most affected individuals have normal parents (if relatively rare)
 - equal distribution between sexes
 - often in consanguineous marriages
 - when one parent and a child is affected $\frac{1}{2}$ of children are affected
- Autosomal Dominant** (p 69, 70)
- * **does not skip generation** (except with poor penetrance)
 - affected person x normal yields $\frac{1}{2}$ affected children
 - distribution between sexes equal
- Sex-linked recessive** (p 71, 72)
- * mothers of affected males usually known to have male relatives affected
 - * sons of affected mothers **all** affected
 - most affected persons are male
 - affected females **all have** affected fathers, mothers at least carriers
 - $\frac{1}{2}$ sons of carrier mothers affected
- Sex-linked dominant** (p 73)
- * affected males **all have** affected mothers
 - * **all** daughters of affected father are affected, not sons
 - does not skip generations
 - $\frac{1}{2}$ children of affected mother affected
 - affected females come from either father or mother

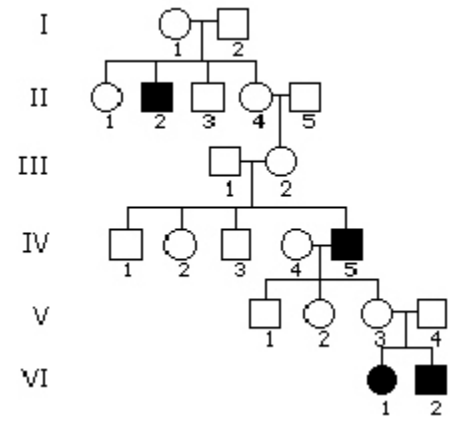
HUMAN TRAITS (alphabetically) (GMSLG 7th p 426, Klug and Cummings, 4th: p 69, Tamarin 4th: p 82-87):

RECESSIVE	affected:	DOMINANT	affected:
albinism	9/10,000	achondroplasia	1/40,000
alkaptonuria	1/25,000	brachydactyly	3.4% to 21%
ataxia telangiectasia	5/ 10 ⁶	Brown eyes (depends heavily on population)	~50+%
color blindness	1/8 ♂	Congenital stationary night blindness (X linked)	
cystic fibrosis	1/1700	Detached earlobe	80%
Duchenne muscular dystrophy	1/3500 ♂	Ehler-Danlos syndrome	1/10,000
galactosemia	1/7,500	Fascio-scapulo-humeral muscular dystrophy	1/20,000
hemophilia A	1/5,000	Huntington disease	7/100,000
Lesch-Nyhan syndrome	1/380,000 ♂	Hypercholesterolemia	1/500
phenylketonuria	1/15,000	Marfan Syndrome	1/5,000
sickle-cell anemia	1/500 blacks	Middigital hair	70% in US
Tay-Sachs disease	1/900 Ashk.Jews	Neurofibromatosis	1/2,750
(1 in 30 Ashkenazi Jews is a carrier)		Phenylthiocarbamide tasting (PTC)	70%
(general population 1/900 is a carrier)		Tongue roller	65%
		Widow's peak	30%

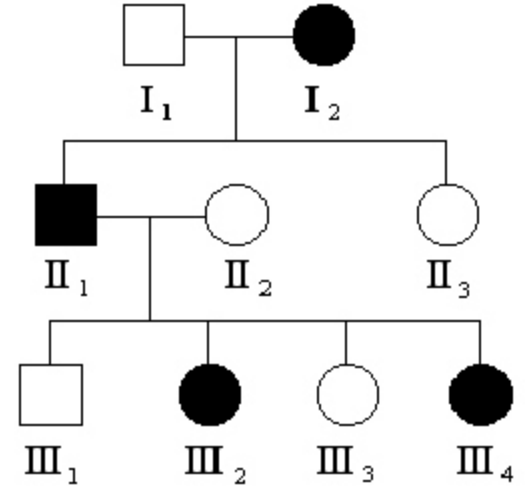
EXAMPLES OF FOUR INHERITANCE PATTERNS

10 January 2011

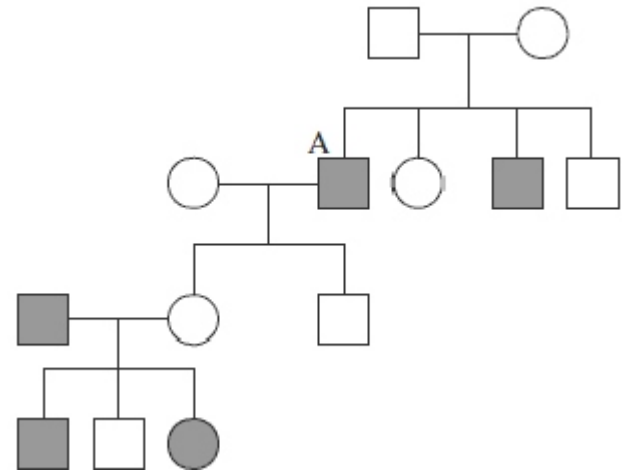
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