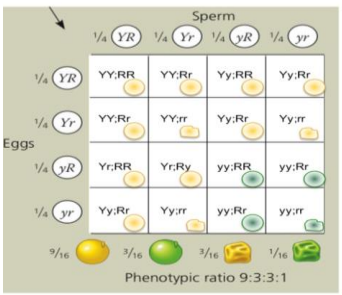
# BMS2042 Elli's lecture summaries; Week 1, Lects 1-3, Autosomal, Sex-linked Inheritance and Extensions

## Lect 1 (Autosomal Inheritance)

**Mendelian Inheritance**: Is used to demonstrate autosomal inheritance, originally by crossing pea-plant species w/ binary traits to see which were dominant or recessive. Mendel bred round (P1) & wrinkled seeds (P2) in 1st gen, & progeny were all round (F1). When F1 interbred, 2nd gen (F2) were ¾ round & ¼ wrinkled. This was the case w/ many other traits, w/ phenotypes disappearing in F1 & reappearing in F2. This led to his 1st law.
**Law of Segregation**: 2 alleles (traits) segregate from each other into gametes w/ each gamete having ½ chance of inheriting each allele, combining at random to form next gen.
**Dominant alleles**: alleles passed down which when heterozygous, phenotype will exhibit dominant's traits.
**Recessive alleles**: alleles passed down which when heterozygous; phenotype won't exhibit recessive's traits.
**Test Crosses**: can establish whether plants are hetero/homozygous for a dominant allele. You cross dominant hetero/homozygous w/ recessive homozygous. If progeny all display dominant phenotype, then unknown is homozygous, if there's ½ dominant & ½ recessive, unknown is heterozygous.
**Dihybrid cross**: can establish crosses of 2 traits at a time. If F1 has 2 dominant phenotypes but is heterozygous, F2 will have a 9:3:3:1 ratio. A 4x4 punnet square establishes genotypes/phenotypes

Yy;Rr x Yy;Rr




Following genotypes are: Yellow + round = Y_;R_. Yellow + wrinkled Y_;rr. Green + round yy:R_. Green + wrinkled yy;rr.
**Autosomal recessive**: males + females equally affected. Skips generations. Common for parents to be related.
**Autosomal dominant**: males + females equally affected. Affected individuals in multiple generations. Transmission by both sexes to both sexes.

## Lect 2 (Sex-linked Inheritance)

**Sex-linked inheritance**: some traits are found on sex chromosomes, like eye colour for Drosophila.
**Reciprocal crosses**: Thomas Hunt bred white eyed female flies w/ normal flies to discover all male progeny were white-eyed & all female displayed wild type. When white-eyed males & wild-type females bred, all progeny displayed wild-type phenotype. This led to conclusion that some genes are carried on sex chromosomes.
**X-linked recessive**: males usually only affected as they have 1X, if X mutated, then males guaranteed to have trait. Females usually carriers as they receive a mutated X from fathers. Affected females will have an affected father and a mother carrier. Male to male transmission is impossible. Includes diseases like haemophilia & red-green colour blindness.
**X-linked dominant**: much rarer w/ both sexes affected (females more so than males). Females are less severely affected than males due to X inactivation w/ affected males transmitting to daughters but not to sons.
**Meiosis**: explains Mendelian Inheritance cellularly. 2 alleles for a gene carried on 2 different homologous chromosomes. Alleles of genes segregate equally into gametes like members of homologous chromosome pair. Different genes act independently as do different pairs of chromosomes. Specific traits are transmitted w/ specific chromosomes.
**Mammals**: have homogametic females (XX) and heterogametic males (XY). This is reversed in birds & reptiles.
**Probability**: used in genetic counselling.
**Product rule**: probability of independent events occurring together is product of probabilities of individual events. In 2 heterozygous parents for cystic fibrosis, probability baby is affected is 1/4. Probability baby is affected then 2nd unaffected baby is ¼ x ¾ = 3/16.
**Sum rule**: probability of either of 2 mutually exclusive events occurring is sum of individual probabilities. 2 heterozygous parents for cystic fibrosis, probability baby is homozygous is 1/2.

## Lect 3 (Extensions to Mendelian Inheritance)

**Different types of dominance**:
**Complete dominance**: if red is dominant, then red + white = red.
**Incomplete dominance**: both alleles aren't completely dominant to each other & blend, red + white = pink.
**Codominance**: both alleles are dominant to eachother, expressed simultaneously. Red + white = red + white (striped)
**Over-dominance**: sometimes, being heterozygous is better for survival than either homozygous. Homozygotes for sickle-cell anaemia will have disease, but also be protected from malaria, if you're heterozygous, you're both protected, & don't have the disease.
**Multiple Alleles**: genes found w/ 3 or more different alleles.
**Polymorphic alleles**: multiple alleles equivalent to wild type, (blood type). ABO system has O which is recessive, A & B which are dominant to O & codominant to each other. All individuals have O surface antigen on RBC surface, w/ ABO locus coding for glycosyl-transferase enzyme adding sugars to normal antigen. O has no enzyme activity & no extra antigens, w/ A & B adding their own respective transferases to make A or B antigens. O, A, B & AB are polymorphic alleles as neither are necessarily mutants, just variants of wild type.
**Monomorphic alleles**: 1 wild type, rest are mutants.
**Lethal Alleles**: An allele which is fatal as a homozygote, recessive & inherited. 2 heterozygous parents for a lethal allele will have a 2:1 progeny, w/ 1 being wild type homozygous.
**Pleiotropic genes**: single genes affecting multiple traits.
**Penetrance**: proportion of individuals of a specific genotype revealing expected phenotype. If they have the allele, what are the odds they'll express it. E.g. 80% of people w/ disease mutation for the dominant disorder retinoblastoma develop retinal tumours. Penetrance is binary, it either happens or it doesn't.
**Expressivity**: degree of expression or severity of phenotype in individuals of specific genotype. Expressivity must be penetrant, but expresses differently.
**Effect of environment**: environment affecting phenotypic expression of genotype. Arctic foxes in winter are white, but in summer are brown due to temperature affecting expression. Humans w/ phenylketonuria (PKU) can't metabolise phenylalanine, but if they remove it from their diet, they won't show symptoms; e.g. environmental chemicals.
**Sex-influenced & sex-limited traits**: gender can govern inheritance pattern for certain traits. An allele is dominant in 1 sex & recessive in the other. Baldness is related to [sex hormone], so males are more bald than females as they have more testosterone, so it's not sex-linked but sex-influenced as father's can pass it to sons. Sometimes traits only occur in only 1 gender (baldness can occur in females if they're homozygous for baldness) called sex-limiting traits. In humans, females will have breasts & males have beards, we have the genes for either, but it's dictated by hormones.