Things to remember in the last hour before the exam: Level 2 Genetic Variation

(This is not a revision sheet – you've done that by now - it's a list of things you might want to remind yourself about …)

1. Cell division
   - Mitosis – for growth / repair – 2 identical diploid (2n) daughter cells – also for asexual reproduction
   - Meiosis –for gamete production (sexual repro.) – 4 haploid (n) cells. NOT identical. When fertilisation occurs makes new individual with full # of genes - maintaining the chromosomal number from generation to generation while promoting genetic diversity and variability within the population.

2. Sources of variation

   Meiosis – formation of gametes – mixes existing alleles into new combinations. During meiosis there is:
   - Crossing over– recombination – pieces of (inward facing) homologous chromosomes are exchanged
     - Recombinants (mix of both parents)
     - Non-recombinants (unaltered by crossing over)

     Genes on same chromosome are linked – don't assort independently – but can be shuffled by crossing over.
   - Independent assortment – each homologous pair of chromosomes lines up at the equator – maternal or paternal –independently of the other homologous pairs
   - Segregation - random which way sister chromatids line up and separate to form gametes

   Mutation - permanent change in the nucleotide sequence in a gene or a chromosome. Mutation is the ultimate source of variation – creating NEW alleles. Change in DNA base sequence → change in amino acid sequence → different protein → potentially different phenotype. Mutations may be
   - Beneficial – improves survivability / reproductive fitness
   - Harmful – decreases survivability / reproductive fitness
   - Neutral – no apparent effect

   Mutations passed on to offspring (heritable) must occur in the gametes →become part of offspring's genetic makeup → transferred to next (& *possibly subsequent*) generations. Mutations in somatic (body) cells affect just that organism – can't be passed on.
3. Changes to gene pool (complete set of unique alleles in a population) – changes in allele frequencies due to
   - mutation (see above)
   - natural selection - survival and reproductive success in individuals whose characteristics are best suited to the environment – at a given time.
     - stabilising – favours middle range of adaptive phenotype
     - directional – favours one extreme of adaptive phenotype
     - disruptive – favours both extremes of adaptive phenotype
   - migration (transfer of genes from one population to another)
     - new alleles being brought in (immigration) or
     - alleles being lost from the population (emigration).

- Genetic drift - change in the relative frequency in which an allele occurs in a population due to random events - not related to the fitness of the allele to that environment.
- Founder effect – very small group leaves much larger group → separate populations – emigrants have only small sample of gene pool – gene freq. is often very different between the 2 populations → decreased genetic diversity in new pop. The small pop will be susceptible to genetic drift.
- Genetic bottleneck – population #s become so low (e.g. many individuals have died) →survivors carry just a proportion of original genes. Inbreeding inevitable & pop more prone to genetic drift.

Migration and genetic drift have a big effect in a SMALL population - relatively small changes in allele numbers can have a bigger impact on the ratio of those alleles in the population

Barriers to gene flow can be geographic – potentially leading to isolation → speciation.

4. Monohybrid and dihybrid inheritance patterns
   - Gene – fundamental unit of inheritance – codes for a protein
   - Alleles – alternative forms of a gene e.g brown eyes / blue eyes
   - Genotype – combination of alleles / genetic make up
   - Phenotype – characteristic coded for by gene –anatomical, physiological, biochemical
   - Dominant – will show in phenotype of present e.g. RR (homozygous dominant) and Rr (heterozygous)
   - Recessive – only expressed if homozygous recessive.g. rr
   - Incomplete dominance - one allele for a specific trait is not completely dominant over the other allele → an intermediate phenotype results from the partial influence of both alleles e.g. snapdragon red + white flowers with pink coloured offspring
   - Codominance - both alleles in a heterozygous organism contribute to phenotype - equally and independently expressed e.g. roan cow has mix of red and white hairs (not 100% "pink" hairs).
   - Sex-linked – genes on X chromosome that are absent on (smaller) Y chromosome e.g. colour blindness , haemophilia. (Other 22 pairs of chromosomes are called autosomes).
   - Monohybrid crosses – involve one pair of contrasting traits e.g black / brown fur. Offspring of cross = F_1 generation, "grandchildren" = F_2. Offspring ratios become closer to theoretical Punnet square ratios when large # of offspring is produced. Bb x Bb give 1:2:1 genotype ratio, 3:1 phenotype ratio.
   - Lethal allele - allele causing death of homozygous individual (allele can be dominant or recessive). Characterised by 2:1 phenotype ratio, rather than 3:1 . No/stumpy tail in Manx cat is result of a dominant mutation, mutant allele M. MM die prior to birth /die young. ~~MM~~ Mm Mm mm 2 no/stumpy tail : 1 normal tail
   - Calculate expected proportions of genotype and phenotype - express as ratio, fraction, % or decimal.
   - To decide if an individual is homozygous or heterozygous do a test cross: breed with a homozygous recessive individual.
   - Dihybrid crosses – 2 pairs of contrasting traits considered simultaneously. Aa Bb can produce 4 types of egg / 4 kinds of sperm → 16 different ways they can combine. Some give the same result so there are 9 genotypes. Hint: Write them in order AB Ab aB ab in punnet square to clearly show proportions of the 4 phenotypes – 9:3:3:1 phenotypic ratio.
   - AABB or AABb or AaBB or AaBb will all have same phenotype.
   - Dihybrid test cross: cross with aa bb. May need to test cross offspring multiple times to be sure.
   - True / pure breeding – homozygous individuals; when mated with own type for several generations, offspring resemble original parents.
   - Multiple alleles e.g. blood types. I^A, I^B and I^O. Group A is I^AI^A or I^AI^O