- Discrete characters don't have to be binary, and they don't have to be equally represented in the population. Traits can also be continuous like height and body mass- continuous characters usually controlled by many genes (polygenic) and/or have strong environmental component
- DNA transcribed to RNA which then translated to protein, how genotype expressed as phenotype
- One gene, one trait
- One gene, many phenotypic effects (pleiotropy). Antagonistic pleiotropy is the idea that the multiple effects of a gene could be antagonistic in their effect – ageing which is beneficial in early life but harmful in later life
- One trait, many genes
- Different genotypes vary I how they respond to the environment phenotype plasticity depending on environment
- Example = Human BMI is increasing which could have an environmental cause (an easier availability to quick calories and having more sedimentary jobs). But individuals' BMI does differ which could show that some people's genotypes handle the environmental change differently or they don't experience the change in environment.
- The central dogma proposed by Francis Crick- the unidirectional flow of information from genotype to phenotype (DNA to RNA to proteins) Changes in proteins cannot affect the DNA (shows how Lamarck's theory of evolution is wrong as he believed physical characteristics acquired during an individual's lifetime e.g.- muscle mass must be passed on through DNA to progeny) not everything in central dogma is correct – for example epigenetic inheritance which means things outside of DNA have power to affect phenotype and are inherited these are referred to as transgenerational effects they are inherited chemical changes to the DNA - e.g. methylation and histones which turn on/off gene expression. This inherited epigenetics can alter the phenotype without altering the DNA sequence - phenotype is affected as sections of DNA can be either turned on or off
- Mutations can change the genotype and may have phenotypic consequences but not always
- Dominant and recessive alleles differ in their affect at a diploid locus (e.g. Mendel's peas)

Changes to the genotype: (1.3 Mark viney)

- Sources of change include mutation changing DNA and the recombination of alleles during meiosis
- Substitution, duplication, and deletion of base pairs
- Transition (A turns into G) when base change from a purine to the other purine or a pyrimidine to another pyrimidine- the most common mutation with small phenotypic consequence
- Transversion (A turns into T) when bases change from a purine to a pyrimidine or a pyrimidine to a purine – has a larger phenotypic consequence
- Cytosine and thymine are pyrimidines, guanine and adenine are purines
- Substitution, duplication, and deletion are caused by errors in copying DNA before cell division- the proof reading is imperfect, so errors do occur
- Mutations can have a small, large or no effect at all it can either be advantageous or deleterious meaning either a loss or gain of function experience sometimes may be lethal it all depends on the precision and where the mutation is.
- A mutations effects on the amino acid depends on what the DNA mutation is and where in the codon it is.
- Synonymous (silent)mutations result in no change, nonsynonymous (missense) mutations change amino acid sequence
- Synonymous mutations are neutral for natural selection whereas non-synonymous mutations are subject to selection
- Only mutations in the germline are inherited- the cells that give rise to gametes (egg/sperm)
- Accumulate somatic tissue mutations with age like cancer. Mutations in germline tissue are what contribute to evolution as they are passed on generations
- 1 human gamete has 3 mutations and one zygote 6 mutations as there are 700.000 UK births per year 4.2 x 10^6 new UK mutations born each year. 76% originate from father as more gametes produced by father – rate of mutation in children increases in parental age

- External agents like radiation and UV can cause mutations too
- Larger-scale chromosome effects also caused by errors during cell division- deletion, duplication, inversion, translocation. The effects depend on the loci affected. Deletion results in loss of genes, duplication means double expression, there will also be errors in mitosis and meiotic recombination which are associated with some human diseases
- Humans are diploid own 2 copies of each autosome meaning we have 2 alleles at a locus, errors in cell division can alter thus e.g., downs syndrome which is the trisomy of chromosome 21. The trisomy of chromosome 21 is a result of non-disjunction during cell division, the separation of chromosomes during mitosis doesn't occur properly
- Mammals differ in X chromosome dose (XX females, XY males) In females one X chromosome is randomly silenced (inactivated) to equalise the X dose between XX and XY- same level of X chromosome gene expression in males and females. Females are mosaic for inactivated for X chromosome

Transmission of the genotype and consequences for the phenotype: (1.4 Mark Viney)

- Cells divide via meiosis and mitosis
- Mitotic cell division products are genetically identical cells this is how embryos develop as it results in somatic tissue growth, produces 2 cells
- Meiosis halves the chromosome number (ploidy) so produces haploid gametes. Produces 4 cells, homologous chromosomes pair which there is a recombination between homologues.
- Sexual reproduction - Diploid Parents undergo meiosis to produce haploid gametes, these gametes then fuse (fertilization) to produce a diploid zygote. This zygote then grows via mitosis.
- Haploids produce gametes via mitosis to preserve ploidy, these haploid gametes fuse to form a diploid zygote which then goes onto grow via meiosis which halves the chromosome number back to haploid
- MEIOSIS……. Interphase DNA already replicated so each chromosome has a sister chromatid attached at centromere. In prophase 1 chromosomes condense and non-sister chromatids cross over and swap DNA. During the cross over chiasmata are formed which hold together the homologous chromosomes. In metaphase 1 homologous chromosomes align at the equator of the cell. Anaphase 1 sister homologous pairs are separated and pulled to opposite ends of the cell due to kinetochore microtubules shortening. In telophase 1 homologues form clusters at either end of the cell, nuclear envelopes reform and cytokinesis occurs resulting in 2 daughter diploid cells each with 2 chromosomes rather than 4, each chromosome contains 2 sister chromatids which are no longer identical due to crossing over. Prophase 2 after a brief interphase new spindles form in each cell. Metaphase 2 chromosomes consisting of their sister chromatids align in the equator once again by their centromere. kinetochore microtubules from opposite poles of the cell attach to each sister chromatid. Anaphase 2 kinetochore microtubules shorten pulling the sister chromatids to opposite poles of the cell. Telophase 2 nuclear membranes reform around 4 clusters of chromosomes cytokinesis results in 4 genetically different haploid cells due to crossing over and random assortment of chromosomes.
- Recombination is more likely between physically distant loci and is less likely at chromosome ends
- LINKAGE = linked alleles are inherited together. strong natural selection acting on one phenotypic trait controlled by one locus means the alleles at that locus will increase in frequency in the population, therefore its linked alleles will also increase in frequency
- In sexually reproducing species the whole unchanged parental genotype isn't transmitted it's a recombined half by each parent that is inherited
- Agametic (without gametes) organisms undergo budding or fission, offspring genetically identical as it is a mitotic process. Parthenogenesis, gametes either produced through mitosis or a modified meiosis, ploidy is maintained offspring identical to parents and each other
- Prokaryotes are hugely abundant and don't have chromosomes instead of circular genomes generally grow by mitosis, conjugation is transfer of genetic material.

- **Punctuated equilibria (1.11 Greg Hurst):**
- Darwin saw evolution as a gradual accumulation of changes over long periods of time. The mutation rate is more or less constant, but the rate of macroevolution is not.
- Darwin and gradualism = evolution progresses gradually, accumulation of small changes over a long period of time results in large differences, he was wrong about macroevolution
- Whereas Eldredge and Gould's punctuated equilibria theory suggests from fossil records that species histories are characterized by long periods of stasis with sudden changes into new forms. Rapid changes are punctuations and stasis are evolutionary equilibrium
- What creates punctuations? 1) plate tectonics and evolution= rearrangement of land over time due to continental drift. Continental movement alters climate as ocean/land interface impacts on climate whereas the center of continent s commonly arid, changes number of shallow seas. It moves areas to new climatic regions for example Australia moved from pole towards the tropics and the UK moved from the tropics to the pole. Taxa are split apart (vicariance). The isthmus of Panama closure 3Mya saw most land species moving north to south. 41% of south American mammals have evolutionary origin in the north.
- 2) mass extinction events and their causes. Mass extinctions don't hit evenly. The Permian-Triassic mass extinction 250Mya the drop in sessile species was a lot bigger. Megafaunas are more likely to go extinct too due to low reproductive rates, slow maturation and low population densities meaning they recover very slowly from periods of high mortality or habitat fragmentation. What causes mass extinctions? Volcanic activity as puts lots of soot in atmosphere which blocks light, large scale climate shifts cause ice caps to melt and sea levels to rise, and single events like meteorite impacts.
- 3) biological innovations= evolutionary novelty alters balance in forms, amphibia were dominant until the reptile form evolved and flying insects were dominant until birds. The early evolution of vertebrates was enabled by genome duplication

Major transitions in evolution 1 (Mark Vine - 1.13/ 1.14)

- Eight major transitions of evolution.

1. Replicating molecules -> molecules in compartments.
2. Independent replicators –> chromosomes.
3. RNA as gene and enzyme –> DNA and protein (code).
4. Prokaryotes –> eukaryotes.
5. Asexual clones –> sexual reproduction.
6. Protists –> multicellularity.
7. Solitary individuals –> colonies.
8. Primate societies –> Human societies with language.

- Why are they 'major'?; Focus on major changes in how information is transmitted between generations. Not major phenotypic transitions. These transitions had to be selected for. These transitions increased fitness. These differences we see now, may not be the initial reason that selection favoured transition.
- **Transition 1** (replicating molecules -> molecules in compartments): Self replicating molecules in the first instance of life, thought to be RNA which is also catalytic (facilitate chemical reactions) , initially individual free "floating" molecules.
  Confining these molecules: concentrates them and other chemicals, so speeding up reactions. Protects the replicating molecules. Keeps the products of replication in the same space. Keeps together mutually beneficial RNA molecules . A COMPARTMENT need not be a cell, but could be molecules absorbed onto a surface.
- **Transition 2 (** independent replicator -> chromosomes): independent replicating molecules (transition 1) = genes……. Chromosomes – collection of multiple genes.
  Chromosomes are slower to replicate. So potentially disadvantageous compared to independent replicators. Theoretically, chromosomes selected for when combinations of genes are needed for