Things to remember in the last hour before the exam: Level 2 Demonstrate understanding of gene expression

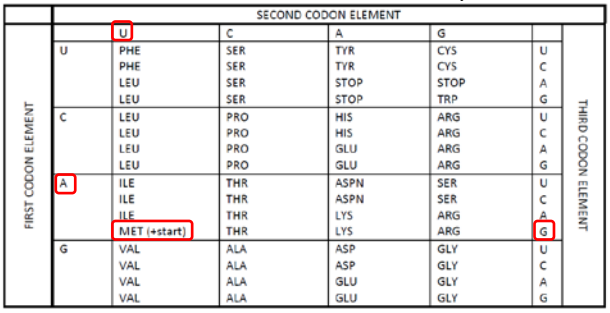
(This is not a revision sheet – you've done that by now - it's a list of things you might want to remind yourself about …)

1. DNA is a polymer, subunits= nucleotides. Nucleotide = phosphate group + deoxyribose sugar molecule + nitrogenous base (adenine, thymine, guanine or cytosine).
2. Two polynucleotide chains are arranged as a double helix, with hydrogen bonds (weak bonds, easily broken) linking pairs of complementary bases (A=T and C≡G).

   E.g. TTGCATGCCATG
   AACGTACGGTAC
3. Proteins are the products of gene expression: DNA → mRNA → polypeptide or protein
4. The information for making a polypeptide/protein is contained in the base sequence of a gene (DNA segment).
5. Proteins are significant because of their structural and catalytic role in living things.
6. RNA.❶ Has ribose sugar instead of deoxyribose ❷ Thymine base (T) replaced by uracil (U).❸ Single stranded.
   - mRNA – acts as copy of a gene and takes the information into the cytoplasm.
   - tRNA – brings amino acids to the ribosomes (the site of protein synthesis).
7. Protein synthesis consists in 2 main stages:
   - Transcription – means COPYING – mRNA transcript (copy) made of one of the DNA strands (template).

DNA	{	T	A	C	C	G	T	C	T	A	A	G	A	Strand 1
		A	T	G	G	C	A	G	A	T	T	C	T	Strand 2
mRNA		A	U	G	G	C	A	G	A	U	U	C	U	

     Strand 1 here is template DNA. As there is no thymine (T) in RNA, uracil (U) bases pairs with adenine (A) so the pairing rules for DNA to mRNA are A=U and C≡G.
   - Translation – information in mRNA is DECODED to create a functional protein. (1) mRNA leaves nucleus (via pores). (2) mRNA forms a complex with a ribosome (usually on ER). (3) Ribosomes move along the mRNA, a codon (3 bases) at a time and 'read' the mRNA bases, in a 5' to 3' direction. (4) tRNA brings in amino acids – (a different kind of tRNA for each of the 20 different amino acids). Three unpaired bases on the tRNA (anticodon) are complementary to the codon on the mRNA. These codon–anticodon 'matches' bring the correct amino acid to the next part of the sequence. (5) Peptide bonds form between amino acids building a polypeptide chain. A start codon initiates the translation. A stop codon ends translation. (6) The polypeptide chain is released from the ribosome and 'folds' into a 3D structure, becoming a functional protein.



SECOND CODON ELEMENT					
FIRST CODON ELEMENT	U	C	A	G	THIRD CODON ELEMENT
U	PHE	SER	TYR	CYS	U
	PHE	SER	TYR	CYS	C
	LEU	SER	STOP	STOP	A
	LEU	SER	STOP	TRP	G
C	LEU	PRO	HIS	ARG	U
	LEU	PRO	HIS	ARG	C
	LEU	PRO	GLU	ARG	A
	LEU	PRO	GLU	ARG	G
A	ILE	THR	ASPN	SER	U
	ILE	THR	ASPN	SER	C
	ILE	THR	LYS	ARG	A
	MET (+start)	THR	LYS	ARG	G
G	VAL	ALA	ASP	GLY	U
	VAL	ALA	ASP	GLY	C
	VAL	ALA	GLU	GLY	A
	VAL	ALA	GLU	GLY	G

A U G	G C A	G A U	U C U
MET (+start)	ALA	ASP	SER

8. The precise sequence of the amino acids determines the properties of the protein made.
9. The genetic code has redundancy - two or more codons can specify the same amino acid. This is known as degeneracy. E.g. GCU and GCC both code for ALA (alanine). As there are more codons than amino acids some codons will be redundant. Advantage of redundancy - when mutations occur, for many codons, a mutation in third base causes NO change in amino acid it codes for. (64 codons for 20 amino acids).
10. Gene-gene interactions. There are a number of possible combinations to the way in which a condition e.g. flower colour, can be inherited because a metabolic pathway has at least two points where different genes control the outcome.
11. "One gene-one enzyme hypothesis" proposed that one gene is responsible for the production of a particular enzyme. Hypothesis has been modified as genes code for proteins that are not just enzymes and many protein molecules are made from more than one polypeptide strand, coded for by different genes.

12. A gene mutation is a change in base sequence of a gene (do NOT write "change in genetic code") resulting in a new allele. This can occur as error during replication (spontaneous) or as a result of a mutagen (induced). They occur randomly, are relatively rare but the majority are harmful. Mutations persist unchanged over successive generations. Mutations occurring in somatic (body) cells can't be inherited.
13. Mutagen – a physical or chemical agent e.g. radiation, uv light, toxin, virus that changes organisms DNA - increasing mutation frequency (above natural background level). Mutations alter genotype. Can be a gene mutation (change in BASE sequence of gene – causing a new allele) or a chromosome mutation. In both cases the genotype is changed.
14. Gene mutations include (1) Base substitution: one base is substituted for a different base E.g. TCA becomes TGA. (2) Frame shifts - base is inserted / deleted, changing reading frame of each triplet code, and altering 'meaning' of the bases that follow.
15. Biochemical reactions are catalysed by specific enzymes and every enzyme is coded for by a specific gene(s). Biochemical reactions form part of a chain reaction so that the product of one becomes the substrate of another step in metabolism
16. A metabolic pathway is a series of biochemical reactions connected by their intermediates: The reactants (or substrates) of one reaction are the products of the previous one, and so on. Because there are a series of biochemical reactions, each one usually controlled by an enzyme, there are multiple places where the end result can be affected. As with most metabolic pathways in our body, the first compound in a pathway is converted to the next compound by the action of an enzyme. E.g. in the pathway A → B → C, the conversion of compound A to B occurs because of the action of enzyme 1, and the conversion of B to C because of enzyme 2. If a mutation occurs which stops the function of enzyme 1, then the end product cannot be made even if enzyme 2 is fully functional. In another individual, a mutation can occur which stops the function of enzyme 2, & the end product cannot be made even if enzyme 1 is fully functional.
17. "Metabolic block". Example. Let * mean an inactive enzyme as a result of the gene: if purple colour is made in two steps then if either of the genes is defective the flowers will be white.

● Colourless raw material -- **Gene A**⟶ colourless intermediate --**Gene B**⟶ purple product =purple flowers

- Colourless raw material -- ***Gene a** ⟶ no colourless intermediate --**Gene B**⟶ no purple product.
  - As there is no colourless intermediate the enzyme coded for by gene B can't make purple product so white flowers.
- Colourless raw material --**Gene A**⟶ colourless intermediate --***Gene b**⟶ no purple product.
  - As the enzyme that would convert the colourless intermediate into purple product is defective no purple product can be made, so white flowers.

aaBB (white flower) x AAbb (white flower) would produce gametes aB x Ab = offspring all AaBb (purple flowers).

18. Phenotype is determined by the presence, absence, or amount of specific metabolic products. There are a number of possible combinations to the way in which a condition can be inherited because the metabolic pathway has at least two points where different genes control the outcome. E.g. If two parents have a disorder such as porphyria, both must have alleles, which result in a deficiency of one of the enzymes at one of the points of the pathway. Normal children can be born, because if the points which are affected in the parents are different, then each one of those can be dominated by an allele inherited from the other, resulting in normal enzyme production.
19. The phenotype of an organism results from the interaction between its genotype AND environment. Environmental factors can include temperature, wind, salinity, available nutrients etc. It is a composite of the characteristics shown by the cell/organism under a particular set of environmental conditions.
20. Genotype provides the instruction set for a particular structure or function, but this may not be fully expressed if the environmental conditions work against it. E.g. plant height is controlled by both genotype and environment. A cutting taken from a plant growing close to the ground due to wind (an environmental factor) can express its genotype fully if grown in a sheltered spot, growing as a tall plant. A plant can't reach its full potential height if nutrients are insufficient to support its growth. Or plant height depends on light availability. The genotype of the plant has not changed at all.