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4/13/13
Exam III Review – Genetics

- *Gene: all the sequences of DNA necessary to encode in RNA, passed from generation to generation, and is capable of change*

Vocab:

- Tautomer- Change in normal base pairing occur with tautomeric shifts (A*-C, C*-A, G*-T, T*-G); are unstable and typically revert back to the original more stable form, A=T and G triple bonded to C
- Aneuploidy- Inappropriate number of chromosomes in a cell
- Inversion- When genes are exchanged between chromosomes during translocation, inversion can be lethal because all of the genes are present yet in an incorrect order, which can clearly lead to mutations; ABCD-> ACBD
- Gene- All sequences of DNA needed to encode an RNA that is capable of change and passes on from generation to generation
- Episome- Plasmid that is inserted into a bacterial chromosome through conjugation
- Enhancer- Cis-regulatory sequence that increases the level of transcription regardless of orientation or directionality
- *Homeotic Gene- Genes that determine which parts of the body form what body parts; involved in determining where, when, and how body segments develop in flies
- Maternal Effect Gene- The phenotype of the offspring is determined dependently by the genotype of the mother due to the genes and transcription factors (TSF)
- Pseudogene- Gene sequences which lack a promoter sequence, continuous ORF with remnants of a polyadenylation sequence; dysfunctional relatives of known genes that have lost their protein-coding ability or are otherwise no longer expressed in the cell
- Unequal Crossing Over- Occurs in repetitive sequences during meiosis where genes align incorrectly and may cause deletions/additions of bases

Material:

- 20 amino acids = 4 nucleotides
  - 1 amino acid = 1 nucleotide (4), 1 amino acid = 2 nucleotides (16)
  - Code is insufficient
  - Has to be 1 amino acid = 3 nucleotides (64)
  - Genetic code minimally has to be read in triplet

- Francis Crick
  - Used homopolymeric sequences to recover certain proteins
  - Code is redundant, degenerate, and universal
    - 64 nucleotide specify 20 amino acids, more than one codon that specifies each amino acid
    - Base pairing at the third position is least important for specifying an amino acid – wobble, doesn't hold the same information content

        - Same for all species – humans, viruses, bacteria – a codon always specifies a certain protein

- Amino acid composition
    - Nitrogen bound to two hydrogen (amine group)
    - Central alpha carbon
    - Carboxyl group
    - R group bound to the alpha carbon – chemical identity that bestows that amino acid with specific molecular chemical and physical properties
    - Strings of amino acids compose a protein; a peptide bond is formed between the carboxyl group and the amine of another
    - Amine group on one end: N terminus, carboxyl group on other end: C terminus, sequencing proceeds N to C direction (5' to 3')

- DNA transcribed to RNA, translated to protein
    - Structure of proteins is a string of amino acids with different properties
    - Amino acid sequence determines higher order structure
    - Protein diversity comes from duplication, modularity (specific motifs with specific functions), accumulate mutations, allow them to move around
    - Primary sequence is responsible for higher order sequence, they interact to form the protein structure, domains and motifs are fingerprints, specific features can be identifying (ex. zinc finger, predict that it is a transcription factor because of this structural signal)
    - Be given the template or coding strand and then we would need to give the other (mRNA, protein, other strand), be able to use a coding table

- Needed for making protein
    - mRNA with 5' cap, UTR, coding sequence, UTR, poly A 3' tail
    - Start codon is AUG, methianine
    - Reading frame established correctly by the start codon

- Process
    - Initiation
        - Ribosomal + mRNA assembly, uses complementary base pairing, initiation factors (IF or eIF)
    - Elongation
        - Protein synthesis
        - Elongation factors (EF or eEF)
    - Termination
        - Liberation of protein

- tRNA/Initiation
    - Carries methionine, uses base pairing
    - 20 different ones to match 20 different amino
    - Charged tRNA

- D loop, T loop, anticodon
  - Synthase recognizes these sequences and then will bind to a specific codon
- Stop codons don't specify an amino acid so aren't recognized by tRNA
- Wobble in the third allows for mispairing
- There's flexibility and multiple transfer RNAs can encode for different sequences
- Independent of the last position sometimes, any of those tRNAs will do
- Anticodon can base pair with codon on mRNA, translation of RNA into amino acid
- Delivery system
- Specificity of bases is important for the process
- A number of different tRNAs have different anticodons, must be charged (amino acyl transfer RNA synthase attaches specific amino acid to specific RNA, specific to each amino acid)
- Charged tRNA, initiation factors bring in the small subunit of the ribosome 30s
  - In eukaryotes: initiation factors bind to the cap at 5' end of mRNA, essentially take up space there, where initiation starts is dictated by the spatial relationship between the start codon and 5' cap, sequence that indicates is the start codon, immediately upstream from that is a Kozak sequence and consensus sequence (RGGRGG, R is a purine); the process is dependent upon the geometry and shape of tRNA (positional information) + Kozak sequence (sequence information)
  - In prokaryotes: sequence immediately upstream from the start codon, Shine-Delgarno sequence, serves same purpose as Kozak sequence, orients ribosome to start at a specific place
- Initiation factors bind to 30s, lock in tRNA, the transcription factors dissociate and 50s can now bind
  - Initiation factors bind to 30s, helping put together the complex of tRNA with methionine, mRNA, and establishes appropriate start (base pairs to stabilize, then dissociates, then 50s comes)

- Elongation
- Three components within a ribosome
- A: acceptor, amino acyl site
- P: central region, peptidyl
- E: exit, where empty tRNAs leave
- EF-Tu, a crucial elongation factor is GTP bound
- Moves one in P site, to the A site
- GTP is hydrolyzed to GDP, fuels ratcheting of complex forward
- Filled E site, growing chain at p site, empty A site
- Ejects empty tRNA, new tRNA is brought to the A site, binding of new EF-Tu to cause exit from the E site and allow tRNA at the A site