Week 6: Metabolic Pathways

1. **Diagram and discuss the metabolic pathway for Tay-Sachs and Sandhoff Disease.**

(a) Describe the phenotype, diagnosis and prognosis and look at the frequency of occurrence in various populations.

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Points: [/]

Feedback:

(b) Indicate the biochemical basis for the disease (i.e, the pathway).

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Points: [/]

Feedback:

(c) What is known about the gene/s and proteins involved – chromosomal location, structure of the gene and size, mutations known, protein structure and function.

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Points: [/]

Feedback:

(d) Any gene therapies?

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Points: [/]

Feedback:

2. **Diagram and discuss the metabolic pathway for Lesch-Nyhan Syndrome.**

(a) Describe the phenotype, diagnosis and prognosis and look at the frequency of occurrence in various populations.

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Points: [/]

Feedback:

(b) Indicate the biochemical basis for the disease (i.e, the pathway).

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Points: [/]

Feedback:

(c) What is known about the gene/s and proteins involved – chromosomal location, structure of the gene and size, mutations known, protein structure and function.

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Points: [/]

Feedback:

(d) Any gene therapies?

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Points: [/]

Feedback:

Week 8: Mapping and Genome Organization

1. **A synthesis of the Huntington's Disease story**:

(a) Describe in detail the phenotype of this disorder.

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Points: [/]

Feedback:

(b) Describe in detail the chromosomal location of Huntington's Disease (HD) and how the location of this gene was worked. Give a detailed description of the process of positional cloning

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Points: [/]

Feedback:

(c) Describe in detail what is known about the structure and function of the HD gene.

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Points: [/]

Feedback:

(d) Develop the status of treatments and gene therapies. Be detailed here as well.

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Points: [/]

Feedback:

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