PORPHYRIAS

Porphyrin is an important part of the heme structure. Without it, our body would not be able to utilize oxygen or break down chemicals in the liver. But, can there be too much of a useful thing such as porphyrin? Yes, an accumulation of porphyrin in the body can result in mild to severe diseases.

Porphyrias are a group of disorders caused by deficiencies of enzymes involved in the production of heme. Heme is a chemical compound that contains iron and is the key component of several important proteins such as hemoglobin, myoglobin, and cytochromes. Hemoglobin and myoglobin are responsible for binding oxygen. In the liver, cytochromes metabolize chemicals (drugs and hormones) so they are more easily removed from the body. Normally, porphyrins leave the body through urine or stool.

Eight different enzymes regulate heme production. There are heme precursors produced at different stages of heme production. Ordinarily, the heme intermediates are modified or expelled from the body, but if one of the eight regulatory enzymes is deficient, the heme precursors or pophyrins accumulate in the body. Porphyrins are the group of heme precursors that accumulate if an individual is deficient in one of the eight enzymes.

Porphyrias are a number of different diseases each caused by a specific abnormality in the process of heme production. In general, the enzyme deficiency is due to a genetic defect leading to its corresponding type of pophyria.

There are three common types of porphyrias: porphyria cutanea tarda, acute intermittent porphyria, and erythropoietic protoporphyria; the less common types are: delta-aminolevulinic acid dehydratase deficiency, congenital erythropoietic porphyria, hepatoerythropoietic porphyria, hereditary copropophyria, and variegate porphyria.

Porphyrias have a differing range of symptoms and severity. Symptoms can include: photosensitivity, increased hair growth, red urine, reddish brown teeth, abdominal pain, and liver damage.

Photosensitivity results from porphyrins being deposited in the skin. When skin is exposed to light and oxygen, the porphyrins generate a charged, unstable form of oxygen capable of damaging the skin, causing nerve damage, leading to pain and paralysis.

Congenital erythropoietic porphyria and erythropoietic protoporphyria affect erythroid cells. Usual symptoms include red urine, deposits on teeth turning them reddish brown and photosensitive skin. There is also increased hair growth on the face and extremities.

Acute intermittent porphyria is the most common type that affects the liver. An individual suffering from this may experience abdominal pain, neurological dysfunction, and red urine. Their skin is usually not photosensitive.

Porphyrias are rare diseases. Men and women are equally affected. A diagnosis involves blood tests and urinalysis.

Treatments depend on the type of porphyria. Drugs that are used to treat acute or sudden attacks include: Hematin given through a vein, pain medication, sedatives, and propranolol to control the heart beat. Other treatments are to increase fluids and glucose which reduce the production of porphyrins, phlebotomy-removal of blood, and beta-carotene supplements. People who suffer from porphyria are also told to avoid sunlight, alcohol, drugs that trigger an attack, skin injury, and to eat a high carbohydrate diet.

Evaluation of the traits	
T*he **Focus** of this work is "Developing"*	I see the focus in this paper being on porphyria; it is what the author discusses most, providing details on the various types of porphyrias. Unfortunately, it takes a while to figure this out. The initial few paragraphs attempt to define the "parts" of porphyria, i.e. the porphyrin ring structure of hemes and the enzymes producing it. In effect, these would be the sub-themes that support the main theme. Although there is a general attempt to make the connection, there isn't much development. It is almost like they are rushing through the details that are necessary to clearly explain why porphyria exists. In other words, they don't develop the sub-themes sufficiently to support the theme.
*The **Organization** of this work "Effective"*	I can identify an overall organization to this work; it starts at the molecular level and evolves through the biochemical pathway to the disease and its symptoms, finishing with the treatments. While this pattern could work, the problem here is that the reader is not prepared to logically move from one level to the next (other than indenting for a new paragraph). There are also some arbitrary statements thrown in here and there (see the second to last paragraph).
*The **Voice** of this work "Developing"*	The voice in this paper is strong in that it uses a variety of sentence structures. The variation is enough to make the reading flow. However, there are times when it gets choppy, mainly in the description of porphyria symptoms. Also the use of a rhetorical question in the first paragraph feels awkward. Word choices are generally appropriate, but there is room for improvement (e.g. "the heme intermediates are modified or <u>expelled</u> from the body" – "secreted" would be a better word in this sentence.)
T*he **Convention** of this work is "Effective"*	The biggest fault for convention in this paper is the lack of figures. In describing the "regulatory enzymes" it would be very helpful to have a reaction pathway drawn out, possibly one that shows the connections to the different types of porphyrias. I also wonder if it would be more effective to place the different types of porphyrias into a table and discuss the relationships between them in the text rather than simply providing a series of individual descriptions. I find no obvious mechanical errors.