Patient's Diagnosis Of Tay-Sach; A Case Study

Tay-sachs is a disease that affects about 1 out of 27 of every ashkenazi jew in eastern europe. If you are not eastern european jewish then you have a 1:300 chance of being a carrier of the disease. The disease happens when 2 people who both have the Tay-sachs gene have a child. The chance of that child getting tay sachs is 1:4. The symptoms of Tay-Sachs disease generally causes a failure of development in the early stages of life even though there are some extremely cases of late onset Tay-Sachs disease. The reason I was able to diagnose my patient (patient b) was because of the symptoms presented that consisted mainly of poor development in the child who was in the early stages of development (6 months old). The symptoms presented were seizures, not meeting common level motor skills milestones, delayed developmental and social milestones, and heightened startle response. The tests provided noted that there was a cherry red spot in the macula. Tay-Sachs is not the only disease that causes a failure of developmental processes in the human body so this alone does not prove that the patient has Tay-Sachs. I have diagnosed this patient with Tay-Sachs disease because the combination of the developmental issues, the age of the child, and the cherry red spot in the back of the macula which is present primarily in Tay-Sachs disease, give sufficient enough evidence that this child unfortunately has this debilitating disease.

The body is in constant homeostasis. Homeo- is a prefix that means: “similar” and -stasis is a suffix that means “state of being”. This means that the human body is in a constant similar state of being. When Tay-Sachs is not prevalent in the body, the body produces the gene HEXA is responsible for providing instructions for producing the subunit of an enzyme called beta hexosaminidase a. This in combination with the directly produced alpha hexosaminidase creates a fully functioning enzyme. The body maintains homeostasis by making sure the enzyme keeps the levels of GM2 gangliosides down in the nerve cells in the brain. The enzyme breaks down the GM2 gangliosides in order to keep the fatty lipids out of the nerve cells so they can function properly.

Because the body needs to maintain homeostasis, it needs everything functioning properly. When someone has Tay-Sachs the gene HEXA has a defect on it. This defect is commonly known as Tay-Sachs disease. This defect, as stated previously, fails to give the body instructions on how to make certain enzymes to break down those gangliosides aforementioned. Because of this the enzyme HEXA is no longer in the body and can no longer provide the function of breaking down Gm2 gangliosides. Due to the fact the body can no longer break down the gangliosides, they build up in the brain causing toxic damage to the nerve cells over time. Eventually the nerve cells are so horribly damaged that the child will just die around 5 years old. This is why the body is unable to maintain homeostasis. The body no longer can stay in a constant similar state of being.

Due to the fact that the patient had presented the absence of development physically and mentally, the patient was eastern european jewish (the people who are most susceptible to the disease), and the cherry red spot in the macula, it is safe to say that this child has Tay-Sachs disease. There is no known cure for Tay-Sachs disease. The only thing to do anything to prevent a child from having Tay-Sachs is to get tested and choose not to have children if you and your significant other are both carriers.