

Free essay on rett syndrome

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There are more than 5000 genetic disorders up to date. These genetic disorders are most evident at birth or during the onset of early childhood. This results to admission in different programs in rehabilitation centers, school programs in order to help the victims affected by such conditions. These disorders can primarily affect the body system, muscles and the respiratory system.

One such disorder is the Rett syndrome. Rett Syndrome is a phenotypically distinct progressive X-linked dominant neurodevelopmental disorder that affects females (Volkmar et al, 2007). There are different types of Rett syndrome. These include the classical Rett syndrome, provisional Rett syndrome and the typical Rett syndrome. The classical syndrome occurs on patients with full diagnostic criteria while the provisional syndrome occurs when the patient has few clinical symptoms while the latter occurs at birth. The disorder has symptoms that include mental retardation, ataxia and growth retardation. This is due to failure in postnatal development as a result of mutation in an X linked binding protein, the methyl-CpG-2. The disorder reduces brain growth and the brain's ability to form new synapses. Its early development in affected children is normal with symptoms usually appear during the first part of life. The condition can be detected when a child responds aggressively to light touch of arms, face or legs and withdraws from objects due to light touches, as well as muscle waste (Umphred et al, 2012). Rett syndrome affects the central nervous system, the brain particularly. The condition occurs as a result of mutation that involves nucleotide substitution, inversions or duplication of short segments of DNA. The nervous system connects different organs of the body that are critical in

the normal functioning of an individual.

It consists of sensory organs, the spinal cord and the brain. These organs interact with others to maintain communication and normal interaction between the organs. The brain forms the central organ of the body that coordinates the functioning of other body parts. The brain controls all other voluntary actions and acts as a central organ to other systems through special fabrics called nerves. The nerves carry information to other body systems and sensory organs. In most cases, gene mutation can inhibit the performance of certain proteins. Mutation alters the protein, and as such can result to a medical condition, always referred to as genetic disorders. One of protein in the tissues that cause Retts syndrome is the MECP2 protein. This protein functions to regulate gene activities. The MCEP2 (Methyl Cytosine binding protein 2) protein in the cells within the helps to maintain synapses between neurons and enhances cell to cell communication. It also helps in alternative splicing that is important in communication between neurons. Retts syndrome occurs as a result of duplication of the MECP2 gene. This causes delayed development, which results abnormality in some neurons. This irregularity in the normal functioning of the brain causes an abnormal behavior. The process affects learning, and communication abilities in coordination due to deletions and insertions in the gene. In a normal and healthy person's body, the brain controls the skeletal system that supports the whole body.

The organs within the body system work together to maintain body position. This helps initiate controlled movements to support the normal lifestyle of an individual. In instances where the brain cannot function normally, other

system organs within the body system can also be affected.

This is because the brain coordinates all aspects of the body. In the case where the brain affects the somatic nervous system, an individual will most likely have problems in body movement. This is because the system handles other muscle activities that connect other vital organs that lead to other diseases like stroke. The ligaments and cartilages with other tissues that connect the bones to help stabilize the body. In the current times, programs in conjunction with other state laboratories work to research on the MECP protein molecule. Known as MECP2 consortium, carry out gene therapy studies on mice to discover more on the functioning of the Rett protein. This has led to major achievements to try and understand the MeCP2 protein. In other cases, the Rett syndrome research trust has developed research strategies to help in identifying the gene that works together with other genes to form the mutated MECP2 protein. In other cases, one can administer an advanced medical procedure to an individual with this syndrome. Therapeutic and other hydrotherapy techniques help alleviate the state by injecting pleasure and relief to patients who suffer from the disorder. These techniques are effective maintaining skeletal functions in order to prevent stereotypical movements that affect the hands. In other cases, the office of rare diseases research (ORDR) and the National Institute of Child Health and Human Development (NICHD), extensively support clinical research on this disorder. Rett syndrome has no cure. This makes the disease cause permanent disability to patients with the condition. However, there is clinical progress towards developing measures and therapies aimed at reducing its effects on patients. The discovery of the

main gene (MECP2) has helped research laboratories to develop better methods of suppressing the condition based on animal models. Patients with these conditions are always under therapy to help them improve their lifestyles through restoring their normal limbic activities. There is no cure and as such the

best alternative is to use support based approaches like occupational therapy. Other Rett syndrome program by the children's hospital in Boston helps adults and young children to realize their potential. The program utilizes expertise to care for children and provides medication to manage the Retts syndrome symptoms. This can increase their muscle strengths to enable them continue with their normal life activities. For instance, high intake of diets rich in calcium, vitamin D and exposure to sunlight enhance bone strength, which can be used to manage bone density. Because of the prevalence of students who go to schools with Autism Spectrum Disorders, there have been changes in the U. S federal law. Students need to qualify for special education in one of an identified disability category. Autism Spectrum Disorder is in a category called low incidence category by the United States Department of Education. The federal educational legislation on Autism Spectrum Disorder enables students with Rett syndrome to receive special education (Joy, 2008). The evolution of the Individuals with Disability Education Empowerment Act entrenches equity in access of education. The education curriculum integrates and establishes collaboration between general and special education. It states the areas that should be improved in order to support children with disabilities. These areas include implementing research based instructional implementations, employing positive behavioral

support and individualized training. The Individualized Educational Program (IEP) helps students to receive special education services. Another key milestone is in the Combating Autism Act passed by the United States Congress.

Its intent is to support the creation and implementation of support care services for children with ASD. The act also allows the government to fund the Centers for Excellence, to facilitate research aspects of students with Autism Spectrum Disorders.

The research aims to explore the causes, diagnosis and possible cures for the disorder. Other legislative resolutions also intend to set days in which people in New York State can be sensitized on Retts syndrome.

Retts syndrome causes mental problems to individuals attacked by the disease. While females are mostly attacked, the disease has its roots in the males also.

In the clinical history, there is no much progress in finding the cure for Retts syndrome. However, other support based programs such as therapy can be used. The future of curing this condition is still unknown although great strides in budgetary allocation for research purposes. Scientists are still exploring ways of finding the cure of Retts syndrome.

References

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