**Colleague Ella M Post to week 6 discussion 6**

This assignment requires that we choose a disorder of brain development, explain the symptoms, discuss how the diagnosis is made, as well as the neurobiological basis for the disorder, and the current options that are available to treat this type of illnesses.  For the assignment, I have researched brain development disorders, and have selected to focus on “Down syndrome”.  Down syndrome is also referred to as “Trisomy 21.”  According to research, this disorder is found to affect “one in every 700 babies in the United States.”  (ret.d from [www.ndss.org (Links to an external site.)](http://www.ndss.org/))

Discovered by the French physician Jérôme Lejeune in the year 1959, Down syndrome was identified as a “chromosomal condition.”  ([www.ndss.org (Links to an external site.)](http://www.ndss.org/))  Research and studies in medicine and science have since been continuous and ongoing, even yet today to try and comprehend the nature of this disorder.  There are many variables that could explain the existence of individual differences. (Baburamani et al., 2019)

**Symptoms**

Some symptoms of Down syndrome consist of intellectual disability; brain degeneration in adulthood. (Carlson & Birkett, 2017)  Features of Down syndrome can vary from person to person.  A few of the more recognizable features include, but are not limited to:

* flattened face
* small head
* short neck
* short in stature
* short fingers
* small hands and feet”. (org)

This is not a set, etched in stone roll call that is characteristic of all Down syndrome patients, and although  intellectual and developmental problems may persists, the degree to which they are affected may range from moderate to severe.   DS children have are known to have “impaired cognitive functionality.”  (Patterson et al., 2013) Other possible difficulties may be delayed language, as well as long-term memory.  (mayoclinic.org) Down syndrome is not inherited but is said to “a mistake in cell division during early development of the fetus.”.  ([www.mayoclinic.org (Links to an external site.)](http://www.mayoclinic.org/))

**How is it diagnosed?**

Screening and diagnostic testing is offered to expectant women and is recommended by the American College of Obstetricians and Gynecologists.  ([www.mayoclinic.org (Links to an external site.)](http://www.mayoclinic.org/)).  The screening process is advantageous in being able to gage the likelihood of the presence of Down syndrome, while the diagnostic testing can provide the results.  (www,mayoclinic.org)

During the first trimester, the test is administered in two parts: the first test which is a blood test will determine the amount of plasma protein-A (PAPP-A) relative to the pregnancy, as well as human chorionic gonadotropin (HCG). This is a pregnancy hormone.  If the presence of both are diagnosed, this may be clear indication of problems with the unborn child.  (www.mayoclinic.org) Next, an ultrasound is given, to inspect certain areas of the neck of the baby, to ensure that the normal amount of fluid has collected in the tissue of the neck area.  If in fact, the fluid has collected in excess, there may be some type of abnormality.

The second semester consists of the close examination of blood levels of pregnancy related substances.  These four substances: alpha fetoprotein, estriol, HCG and inhibin A are related to the pregnancy process.

In some cases, the doctor may order a test known as a “chromosomal karyotype”.  This is done to test the child’s chromosomes.  Should there be found additional chromosome 21 in all or any of the cells, the diagnosis will be Down syndrome. ([www.mayoclinic.com (Links to an external site.)](http://www.mayoclinic.com/))

**Neurobiological basis of Down syndrome**

Down syndrome is a chromosomal abnormality and can be considered as a neurological disorder.  It is like neurological complexities such as epilepsy, strokes, cervical spinal cord compression and basal ganglia damage.  The abnormalities can facilitate “intellectual disability” (Carlson & Birkett, 2017).  In other research studies, MRI history provided data suggests that brain volume within children and youth is smaller, having  decrease in cerebellar and hippocampal masses.  (Gunbey et al., 2017)

**Treatment**

In the study and learning and insight into this debilitating disorder, it is of the essence that appropriate tools be at the disposal of those who are studying this phenomenon.  The study of the brain is no less needed today than it was years ago.  More insight and understanding are needed to be able to assess and evaluate patient needs.  Thus the demand for effective methods of research plays a key role in the detection, treatment and ultimate eradication of any disorders of the brain could mean a decrease in mental health illnesses.  Among techniques hailed as a source that can “ provide information”  (Carbó-Carreté et al., 2020)  It is a non-invasive technique that is useful in the assessment of function of brain activity and is regarded as being highly valuable.  (Carbó-Carreté et al., 2020).

While DS cannot be cured, those with DS are owed efforts for a chance at a better quality of life using future studies and assessments and testing tools.  These resources will provide a deeper insight and understanding of tracked data to help individuals according to their needs.

**Reference:**

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