Down syndrome essay

Down syndrome (usually abbreviated DS) is a medical condition caused by a chromosomal disorder in which the patient has 47 chromosomes instead of the normal 46. People suffering from Down syndrome are characterized by mental retardation (with a mean IQ score of less than 50), dysmorphic facial characteristics, and many other phenotypic features. It is believed that the condition is the most popular genetic cause of mental and intellectual retardation and disability. This paper will analyse the cause, diagnosis, symptoms and the risk factors associated with this condition. It will give a case study, in terms of research, which shows the importance of not having children very late in life, as this may increase the risk of Down syndrome in the children.

Cause

Down syndrome is mostly caused by the abnormal nature of the patient having three distinct copies of chromosome 21(usually called trisomy 21) in each cell of the body. On a few occasions though, Down syndrome may be caused when there is an additional chromosome in some of the body cells, usually referred to as mosaic trisomy 21. In some cases, there may be either an additional copy or portion of chromosome 21 that is attached onto another different chromosome (this is called translocation trisomy 21). Down syndrome affects all organs in the body which lead to organ system malfunctioning and impairments, both cognitive and physical (Pinto & Schub 2011).

Effects

Down syndrome affects the organ system of the body leading to defects such as hearing and visual impairment, celiac disease, congenital heart disease, and autism. It can also lead to the onset of the Alzheimer's disease (Pinto & Schub 2011).

Diagnosis

Down syndrome can be diagnosed at the prenatal stage. Diagnosis can be done using several ways, such as chorionic villus sampling, amniocentesis, as well as sampling the percutaneous umbilical blood. These tests can be invasive in nature though. Additionally, a specialized risk assessment can be conducted to analyse the risk levels and determine if further testing should be done. Finally, the condition can be diagnosed during birth if it had not been detected prenatally. The diagnosis at this stage is done via the help of cytogenetic studies (Pinto & Schub 2011).

Symptoms

Persons with Down syndrome show varying signs accompanied by different medical conditions. Some of the more common physical characteristics of a Down syndrome patient are a flat face and nasal bridge, uncharacteristically short neck, slanted eyes that have epicanthal folds, very little hair (on the head) and eyelashes, very small ears, a crease in the palm (hand), whitish spots on the iris, brachycephaly, and a protruding tongue. Some male patients may have undescended testicles. Infants with the condition may suffer from hypotonicity as well as a seizure disorder (Pinto & Schub 2011).

Down syndrome may also be manifested in persons through hearing and visual impairments, a never-ending constipation, congenital heart disease, hypothyroidism, and endocarditis. Other medical conditions associated with Down syndrome are atlantoaxial instability which can lead to neurological disorders, celiac disease, stenosis, URI’s, atopic dermatitis, insomnia, and obesity. Adults with this condition tend to show signs of aging at an unusually early age. These signs include loss of hair (early greying), wrinkling of the skin and neoplasms. Young adults and adolescents may also suffer from degenerative vascular disease. Infant patients take a longer time to develop their speech and language skills (Pinto & Schub 2011).

Treatment

Persons suffering from Down syndrome may exhibit differing levels of disabilities. In most cases, the patients are allowed to live with their family members where they have the best chance of developing their social well-being. On the other hand, some of them may require a specialized habitat where they will be under intense supervision while being put through some form of individualized routine tasks. This type of patients is said to have higher levels of the disease and thus require more help by experts. The treatment of the patients will also be guided by the level of disability. However the treatment is rather complex due to the many organs that are affected. Therefore, the treatment includes many specialized health care experts from many fields. These health care experts include cardiologists, orthopaedics, neurologists, and ophthalmologists. Therapists dealing with physical and mental well-being, speech and language development and nutrition may be involved as well (Pinto & Schub 2011).

Risk Factor

It has been proven that the risk of a child being born with Down syndrome increases as the maternal age also increases. This simply means that a mother who gives birth at 40 years has a higher chance of getting a child with the condition than a mother of 25 years. For instance, mothers of 35 years have a risk of 1: 385 of giving birth to a child with the condition. This risk increases to an incredible 1: 106 for 40-year-old mothers. The risk is even more for mothers at 45 years of age, at 1: 30. This simply suggests that would-be mothers should not wait until it is too late to have a child, as the children will be at an elevated risk of having Down syndrome (Pinto & Schub 2011).

Case Study

Most couples are always happy when their child is conceived. They are always expectant of getting a healthy baby who would later turn out to be a great person. The anxiety that accompanies the conceiving and birth of a child is always intense. While expecting the best for the unborn child is great indeed, the prospect of getting a very healthy baby is not so assured. That is why couples are always advised to visit the antenatal clinics at different stages of the pregnancy to ensure that the foetus is growing as expected. It is during these visits that any anomalies can be detected and, if possible, rectified. As the paper has already mentioned, Down syndrome can be diagnosed in a child prenatally or at birth. However, it is advisable that the mother goes for the tests very early during the pregnancy. This would make diagnosis easier. Thus, couples should always plan to pay a visit to the

prenatal clinicians so that the foetus is tested for Down syndrome. This should be done early in the pregnancy, most preferably in the first trimester.

Down syndrome is the commonest cause of chromosome disarrangement in the human body. However, a subcutaneous pile up of fluid just behind the neck some eleven to thirteen weeks into the pregnancy (+ 6) can be visualized and scanned by ultrasound as being nuchal translucency. The increase in nuchal translucency in the foetus signifies the increase in the risk of the foetus developing chromosomal anomalies, as well as other foetal disorders and genetic syndromes. This simply suggests that the increase in the nuchal translucency can signify the development of Down syndrome. The following section discusses a case study, in terms of a research, that was conducted to prove why having children at an older age increases the chance of the baby having the condition.

Aim of the Research

This research was primarily aimed at evaluating the nuchal translucency screening for Down syndrome in the foetus (at 11 -13 weeks (+ 6 days)) of the pregnancy in relation to the maternal age. The research was conducted and written by Disdarevic et al., in 2011.

Methodology

In this research, a total of 105 pregnant women were participating. These women were supposed to be between eleven to thirteen weeks (+ 6 days) into the pregnancy at the time of the research. The research itself was conducted for five months at the University of Sarajevo’s Department of

Gynaecology and Obstetrics. The studied mothers had to be strictly within the set gestation period. This had to be confirmed using information from the women’s last menstrual period. To be assured of the credibility of this information, the Crown rump length of the foetuses was measured. At this time, this length should be ranging from about 45 to 84 mm.

Procedure

First and foremost, the foetus is viewed longitudinally (the midline section). This view is then obtained with the corresponding image, magnified in such a way that the foetus will fill all or most of the image. When this has been done, the unborn is measured from the head up to the rump. It should be ascertained the foetus is neutrally positioned. The foetuses were found to be within the rangee of 45 to 84 mm long.

Now that the foetuses are of the prerequisite age, their respective nuchal translucency was thus measured. This was done using five ultrasonographers that conformed to the guideline set up by the Fetal Medicine Foundation. This measurement also strictly followed conventional methodological criteria. The scans were conducted with the help of trans-abdominal ultrasound in the middle section of the sagittal plane. The foetus at this stage is made to cover about three quarters of the image. All the measurements (Callipher) were rounded off to 0. 1 mm. To reduce the occurrence of large errors, a lot of care was taken to ensure that both the skin of the foetus and amniotic membrane were clearly visible. The subcutaneous translucency, being the main object of the research, had its thickness measured not once or twice, but for several times. The largest recorded thickness was the measurement

used in the analysis. There are some cases that required the use of foetal karyotyping. Such cases included those of women that were more than 35 years of age and whose family history has experienced aneuploidy at some point and the cases where the measured nuchal translucency was more than 3 mm.

Results

The median age of the pregnant women in the study was 33. 0 years, the inter quartile range age was 15 years. Out of the 105 women that were studied, 54 of them were 35 years of age or over. This represented 51. 4 % of the total. For the women who were 35 years or over, the nuchal translucency recorded more than 2. 5 mm in six foetuses; 2. 6 mm in four of them and 3. 6, 4. 5 mm in the other two. The 3. 6 mm and 4. 5 mm cases were measured in the pregnant women that were 35 and 37 years old, respectively. Additionally, there was one case where the nuchal translucency was measured to be -4. 5 mm. This simply meant that this foetus had Down syndrome. There was also one karyotype case that was found to be normal, at nuchal translucency -3. 6.

For the pregnant women that were below 35 years old, the nuchal translucency measurement was normal. It was also discovered that there was statistic difference in the frequency of nuchal translucency values of more than 2. 5 mm between the two groups of women.

Discussion

Trisomy 21 comes about due to an anomaly in the spermatozoid or the egg cell. It can also come about when there is a genetic error, while the unborn is still at the embryonic stage. The increasing risk of Down syndrome in babies with increasing maternal age can be attributed to the relatively longer latent period of egg cells of the women at the primary follicle level, coupled with the diplogenetic subphase process of the meiosis (usually referred to as I prophase). This extended period of the egg cell in the latent state may result in disorders in the cell metabolism. As a result, the normal cellular divisions as well as the proper arrangement of the chromosomes would be interfered with. This simply means that haploid formation would be impaired. Lastly, the regular number of chromosomes would not be polyploid (Dizdarevic et al. 2011).

Studies have shown that the increase in nuchal translucency can be used to identify up to 76 % of foetuses having trisomy 21. Additionally, in the 1, 015 foetuses that are currently undergoing karyotyping during the first trimester of the pregnancy due to increasing thickness in the nuchal translucency, it has been proved that the risk of chromosomal disorders increases with maternal age, as well as the nuchal translucency thickness (Nicholaides 2004). Lastly, it has been observed that the incidences of trisomies 21, 18, 13 in the unborns with NT thickness of 3, 4, 5, and more than 6 mm were estimated at 3 times, 18, 28 and 36 times higher than the expected incidences, based on age of the mother (Pandya et al. 1995).

Conclusion

Down syndrome can bring a lot of suffering to the patients and their families. The good thing is that the condition can be detected and diagnosed well in time. This fact underscores the importance of pregnant mothers visiting the clinics regularly, well before the child is born. However, as we have seen, the risk of getting a child with Down syndrome increases with the maternal age. Therefore, it is advisable for couples not to have children after 40. If this is not possible, then they should be constantly in touch with their doctors.