Down Syndrome 3 Essay, Research Paper

Down Syndrome

Down Syndrome is a chromosomal disorder. It occurs in about 1 of every 800 births. People with Down syndrome may have mild to severe learning disabilities. Physical symptoms include a small skull, extra folds of skin under the eyes, and a protruding tongue. People with Down syndrome are subject to a variety of medical problems including heart abnormalities and thyroid gland dysfunction. Survival rates have been increased dramatically in recent years as problems specific to Down syndrome become known, allowing the early treatment. The life expectancy of people with Down syndrome now approaches that of people with out it. Usually it s around 55 years old. You would have numerous abnormalities; it wouldn t go over well in school. People don t accustom to that very well in public schools.

The genetic cause for Down syndrome is when a person inherits all or part of an extra copy of chromosome 21. Trisomy 21, the inheritance of an entire third copy of this chromosome, accounts for 95 percent of Down syndrome cases. Two other abnormalities each account for 2 to 3 percent of all cases. The first, translocation, takes place when a child inherits an extra piece of chromosome 21 attached to a different chromosome. The second, called mosaic Down syndrome, results when only some cells in the body have the extra chromosome. There is no cure for Down syndrome although prenatal tests are available to identify fetuses with the disorder.

Down syndrome can be diagnosed just by looking at the baby at birth. The facial features and characteristics can tell you that. If the attending physician suspects Down syndrome, a karyotype a blood or tissue sample stained to show chromosomes grouped by size, number, and shape will be performed to verify the diagnosis. The most familiar physical traits of Down syndrome include:

+ Low muscle tone

+ Flat facial profile

+ Upward slant to the eyes

+ Abnormal shape and small size of the ears

+ Single deep crease across the center of the palm

+ Excessive ability to extend the joints

+ Fifth finger has one bending joint instead of two

+ Small skin folds on the inner corners of the eyes

+ Excessive space between large and second toe

+ Enlargement of tongue in relationship to size of mouth

The majority of these defects can be corrected, resulting in long-term health improvements. Children with Down syndrome also tend to have increased susceptibility to infection, respiratory problems, obstructed digestive tracts, and childhood leukemia.

With Down syndrome there is no therapy except some of the defects listed above can be corrected like I stated. But the features of the face remain and cannot be fixed unlike the other ones that can be fixed. There would be no therapy in the future that I can find in my research.

Yes, it is possible to detect Down syndrome when the infant is in the uterus. You can do so by screening tests like the Triple Screen and the Alpha-fetoprotein Plus. Both tests measure quantities of various substances in the mother s blood, and together with the woman s age, estimate her risk of having a child with Down syndrome. Typically they are offered between 15 and 20 weeks of pregnancy. More accurate are diagnostic tests, which include chorionic villus sampling, amniocentesis, and percutaneous umbilical blood sampling. While these procedures are about 98% to 99% accurate in their detection of Down syndrome there is an increased risk of miscarriage because these tests are performed inside the uterus. Because of this risk, they are recommended primarily for women over the age of 35.

For genetic counseling, there is no possible way to stop Down syndrome from happening. It s inherited and it just depends on if the chromosome 21 gets a third chromosome. Someone with Down syndrome could have a child and it could be perfectly normal just depends on the count of chromosome 21.

References

www.kidshealth.org (5 pgs.)

Encarta Online Premium (2 pgs.)