Down Syndrome Essay, Research Paper

Down Syndrome

Down Syndrome, which used to be called mongolism, is a congenital

malformation accompanied by moderate to severe mental retardation, is caused

by a chromosomal abnormality.

People with Down syndrome are often short in height and have a small,

round head with a high, flattened forehead and split, dry lips and tongue. A

typical feature is a fold of skin, the epicanthic fold, on either side of the bridge

of the nose. The palms show a single transverse crease and the soles have a

straight crease from the heel to the space between the first and second toes.

These people are also subject to congenital heart defects and tend to develop

leukemia. Their intelligence quotient, IQ, usually ranges between 20 and 60,

but with early intervention and proper education, some people reach higher

levels.

The overall incidence of Down syndrome is approximately one in 700

births, but the risk varies with the age of the mother. The rate of Down

syndrome in children born to 25 year old mothers is approximately 1 in 2000,

but the risk increases to 1 in 200 for 35 year olds and more than 1 in 40 for

women older than 40 years. Prenatal tests such as amniocentesis and chorionic

villus sampling can be used to detect the chromosomal abnormality causing

Down syndrome. In addition, maternal blood tests can suggest the presence of

a fetus with Down syndrome when levels of alphafetoprotein are lower than

usual, or when levels of unpaired estriol and human chorionic gonadotrophin

are abnormal.

The chromosomal abnormality involved in most cases of Down

syndrome is trisomy 21, or the presence of three copies of the 21st

chromosome. As a result, the affected person has 47 chromosomes in all body

cells instead of the normal 46, although how this causes the condition’s

symptoms is not yet known. Scientists assume that the reason for the abnormal

chromosomal combination is the fertilization of an ovum having 24

chromosomes by a sperm with a normal assortment of 23, but they have also

found that the sperm can carry the extra chromosome as well. The abnormal

ovum or sperm is derived from a germ cell in which the pair of 21st

chromosomes holds together and passes into the same sperm or ovum instead

of separating. In the type of Down syndrome called translocation, the extra

chromosome 21 material is attached to one of the other chromosomes; when

some, but not all, of the body’s cells carry an extra chromosome 21, the

condition is a type of Down syndrome called mosaicism.

Down syndrome is not yet responsive to medical treatment, but better

medical care of the accompanying disorders and infections now results in an

almost normal life span instead of the previous life expectancy of about 14

years. The severely retarded may still be institutionalized, but many Down

syndrome children are raised at home, where their full potential can be

developed. Although, with an average mental age of about eight years, they

continue to need a protective environment, many adults with Down syndrome

work in sheltered workshops; some hold regular jobs in business and industry.

Bibliography

www.downs.com

www.americahealth.net