



## Down Syndrome: Its Impact on families

The first physician to identify genetic chromosomal basis of Down syndrome was a British Physician, John Langdon Down. Langdon Down was named after this syndrome because he was the first Guy to identify it as a disability. Sequel to the effort made by Langdon Down, this disorder was identified as chromosome 21 trisomy by Jerome Lejeune, in 1959.

Since after then, approximately 4,000 children with Down syndrome are born in the U.S. each year, or about 1 in every 800 to 1,000 births. Statistics is not very clear of the number of individuals in Africa and the Caribbean living with Down syndrome. What I know for sure is that thousand and millions of people in the United States have Down syndrome, than hundreds of children and adolescents in Africa and the Caribbean which is a chronic health condition or a recurring and persistent condition that affects a person's physical health. More than 90 million people in the United States live with a chronic health condition (Meek, L et al, 2005). Based on mortality trend, one could say that more than 5,000 children and adolescents suffer from this syndrome in Africa and beyond because healthy food and environmental conditions are under question. Down syndrome leads to genetic cause of mild to moderate intellectual and developmental disabilities (NADS, 2010) in children whose families are genetically tied with this disability. The average IQ of people with Down syndrome is about 50, compared to normal children with IQ of about 100 (Liptak, 2008). Scientists in modern times call this disability, chromosomal disorder because it is caused by an error in cell division that results in the presence of an additional three chromosome trisomy, 21.

In any Biology classes, you will come to learn about reproductive organs in human body and how it works to keep human body active. In any study of anatomy, you will also learn that human body is made up of cells and these cells are made of chromosomes. Human cells are made up of 23 pairs of chromosomes, half of which are inherited from each parent. Scientists later identified chromosome pair for male as XY while chromosome pair for female as XX. It is believed that when reproductive cells of sperms and cells of an ovum combine at fertilization the fertilized egg that result would contain 23 chromosome pair. A fertilized egg that develops into a female XX will contain a chromosome pairs 1 through 22 while a fertilized egg that develops into male XY; will contain a chromosome pair 1 through 22. And when the fertilized egg contains extra material from chromosome number 21, this results in Down syndrome disability.

There are of course three genetic conditions that cause Down syndrome in children. These three genetic conditions are caused by the presence of extra-chromosome 21 in all cells of an individual. The extra chromosome in the body originates in the development of either the egg or



## Down Syndrome: Its Impact on families

the sperm as the case maybe. As a result, when the egg or sperm unites to form a fertilized egg, three rather than two chromosomes 21 are present. The extra chromosome is repeated in every cell and this repetition is called trisomy, 21. Genetically speaking, Down syndrome is due to mosaic trisomy, 21. This extra chromosome changes the orderly development of the human body and brain. Therefore, a Down syndrome based on genetic or due to Mosaic trisomy, 21 definitely have 46 chromosomes in some cells. Children who have this problem will equally have 47 chromosomes including an extra chromosome 21 in others. An increase or tendency for Down syndrome in children is as a result of increase in maternal age of parents. Women who become pregnant at age 35 and above have the likelihood of having babies with Down syndrome. The likelihood of having babies with Down syndrome continues to increase as women ages. Advancement in age of parents can cause an event that may occur during formation of reproductive cells, the sperm or ovum. Against these backdrop, Eunice (2010) quipped that there may be increased likelihood of Down syndrome in future pregnancies by parents of a child with Down syndrome due to translocation trisomy 21. This experience is common especially when one of the parents maybe a balanced carrier of the translocation. Again, Down syndrome is a result of mitotic error; an error in cell division which occurs after fertilization when sperms and eggs are joined.

### Characteristics of Down syndrome

Characteristics of people with Down syndrome resulted out of the evolution of genetic revolution. The consequence to any peace at home, family, school and society was rendered ever more fragile by categorizing them as special education students in schools or special people with needs in families and society. Poor muscle tone; Slanting eyes with folds of skin at the inner corners (called epicanthic folds); Hyper flexibility (excessive ability to extend the joints); Short, broad hands with a single crease across the palm on one or both hands; Broad feet with short toes; Flat bridge of the nose; Short, low-set ears; Short neck; Small head; Small oral cavity; and/or Short, high-pitched cries in infancy, all this make them needy individuals in our families (See, Health Newsflash, Online Service). Most patients with Down syndrome are also diagnosed with Multiple Sclerosis. MS is a disease in which the protective covering of nerve fibers in the brain and spinal cord are destroyed. Patients who suffer from Multiple Sclerosis always experience tingling and numbness in their body and they may feel tired and dizzy at all times. There is no cure for MS and this disease is common in young adults than in old people. Treatment of MS therefore, includes but not limited to: physical therapy to strengthen muscles, medications, avoidance of stress and extreme temperatures, psychological counseling, and



support from family and friends.

behavioral problems associated with this disability may include: Impulsive behavior, poor judgment, short attention span and slow learning skills in school etc. Symptoms individual go through could vary from individual to another. Some individuals may have a lower resistance to infections and this condition cause them to develop respiratory problems or sleep apnea. Obesity however, is another tendency that children with Down syndrome go through. There are more than 100 characteristics of Down syndrome in children. Many of them eat break fast before coming to school and still join in the school breakfast and lunch only to return home and have another heavy lunch in a space of four hours. Study shows that individual with Down syndrome suffers from a condition known as Atlantoaxial instability. Atlantoaxial instability is a misalignment of the top two vertebrate of the neck. Misalignment of vertebrate can cause patients to be at increased risk of developing certain medical conditions not necessarily associated with Down syndrome, such as congenital heart disease. Other conditions associated with this syndrome includes but not limited to: a flat facial profile, an upward slant to the eye and learning disabilities etc. Children with Down syndrome suffer from medical health-related conditions.

Some of these conditions include:

Thyroid problems Hearing problems Congenital heart disease Eye problems Seizure disorders Bone, muscle, nerve, or joint problems Leukemia and other cancers Immune system problems Developmental delay Mental retardation Alzheimer's disease (Emed TV, 2006, Clinaero, Inc).

Treatment of Down syndrome

There is no cure for Down syndrome since it is a problem with gene and chromosomes. Drugs or medicines are not used to treat Down syndrome either. However, medicines are used to treat other related diseases associated with Down syndrome and other health problems that may arise or develop as the case maybe. Antibiotics can be used for ear infections and thyroid hormones for under active thyroid gland (hypothyroidism). Treatment of Down syndrome also focuses on controlling symptoms and any medical conditions that come along as a result of Down syndrome. Some medical condition observed on people with Down syndrome requires expert medical surgery. Half the number of people with this problem has congenital heart disease and early onset of pulmonary hypertension (high blood pressure in the lungs). congenital heart problems include: Tetralogy of Fallot Persistent ductus arteriosus Atrial septal defect Ventricular septal defect.



## Down Syndrome: Its Impact on families

Doctors have been warned time without number to engage in initial diagnosis of Down syndrome at the birth of the child based on how such baby looks. Therefore, doctors may hear a heart murmur when listening to Babies chest with stethoscope (Neil, K et al, 2010). Nurses and healthcare providers must have a blood test done to check for extra chromosomes. There are other tests that are required to be done because they are essential, namely: Echocardiogram, ECG and X-rays of the chest and gastrointestinal tracts (Neil, K et al, 2010). Again, counseling is believed to be beneficial to patients and parents at the onset of this syndrome. Meeting with occupational therapists, Nurses, counselors or member of the clergy can be very beneficial in coping with stress associated with this disease. A Down syndrome support group can be very helpful to patients and families in letting out their feelings and concerns (Auther, Schoenstadt, emed TV).

Behavioral training can help people with Down syndrome and their families deal with frustration and Anger and compulsive behavior that may arise in the process. This is why parents and care givers should learn to help a person with Down syndrome deal with frustration and irritability. Special education and training are relevant for children with delayed mental development. Speech therapy would help improve language skills or expressive development. Physical therapy would teach movement skill. Occupational therapy would help with feeding and performing skills. Mental health care would help both parents and the child in question to manage mood or behavior problems (Neil, K. et al, 2010). Women who are at high risk (advanced maternal age, positive AFP tests, or a history of previous child with Down syndrome) may benefit from additional ultrasound scans between 18 and 22 weeks of pregnancy.

Patients and families with Down syndrome disease must ensure that they safeguard patients and guard against their immune systems- i.e. protection of the inside of the body. The immune system, the white blood cells help the body fight pathogens as lymphocytes. When a pathogen enters the body, lymphocytes multiply in lymph tissue to fight any kind of infection. This immune system helps patients with Down syndrome develop lasting immunity. Immunity is the body's resistance to disease due to presence of antibodies. For example, after a patient with Down syndrome recovers from an illness like chicken pox virus, the same chicken pox antibody would remain in the body of such patient and protect him or she from developing the sickness caused by chicken pox again. Patients with Down syndrome therefore must be safeguarded in order for them to develop active immunity. Active immunity is resistance to disease due to the presence of antibodies. After a patient with Down syndrome recovers from chicken pox virus, the chicken pox antibody remains in his or her body and protects him or her from developing



## Down Syndrome: Its Impact on families

chicken pox (Meek, L et al, 2005).

Gerald Ogbuja

More Information on Down syndrome

National Down syndrome Congress 1370 Center Drive, Suite 102 Atlanta, GA 30338 (800) 232-6372; (770) 604-9500 Web address: <http://www.ndsccenter.org/> National Down Syndrome Society 666 Broadway, 8th Floor New York, NY 10012 (212) 460-9330 (1-800) 221-4602 (Toll Free)

Web address: <http://www.ndss.org/>

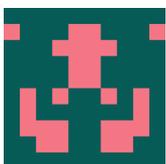
The Arc of the United States (formerly the Association for Retarded Citizens of the United States) 1010 Wayne Avenue, Suite 650 Silver Spring, MD 20910

References National Association for Down syndrome (2010) retrieved November 19, 2010 @ <http://www.nads.org> Eunice K Shilver, (2010), Facts about Down syndrome, National Institute of Child Health & Human Development, National Institute of health. Health Newsflash (2010) Down syndrome facts and Information, retrieved November, 19 2010 @ <http://www.healthnewsflash.com> Meeks, L Heit, P & Page, R (2005), Health and Wellness, the MC Graw-Hill, Glencoe Companies, Inc Neil, K.K & David, Z (2010) Down syndrome, Medline plus, US National Library of Medicine, National Institute of health

Author

Recent Posts

---



Gerald Ogbuja

---