Commentary

# Down Syndrome: An Insight of the Disease

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# DESCRIPTION

Down Syndrome (DS) is a genetic chromosomal disorder, specifically involving a third extra copy of the 21st chromosome. This genetic disorder occurs due to the presence of all or a part of the Extra 21st chromosome, so this way we call it TRISOMY 21 also. Because in this condition the three copies of the 21st chromosome occur within the cell instead of 2. We usually have Two 21st chromosomes, or we can say all 22 autosomes occur in pairs-one from the father and one from the mother. But in the case of Trisomy the 21st chromosome occurs three times [1-4]. Two from one parent and one from the other and mostly in Down's syndrome we get one copy from the father and two copies from the mother. This causes mental retardation and slower physical development. Abnormalities in facial appearance such as slanting of the eyes, smaller than the average skull, and protruding tongue. Kids determined to have the condition have been known to be deficient with regards to muscle tone and have free joints. Down syndrome impacted individuals have additionally been known to foster heart conditions sometimes down the road. Regardless of being hereditary, Down syndrome is seldom acquired. Effects of this disorder-Thyroid problems, Hearing problems, Congenital heart disease, Eye Problems, Bone, muscle, nerve, or joint problems, Leukemia and other cancers, Immune system problems, Development delay, and Mental Retardation Physical Signs of this disorder - Flat face, upward slanted eyes, deep palm creases, loose ligaments, small hands, short neck, and abnormally shaped ears. Down syndrome symptoms are few physiological issues managing Down's syndrome incorporate inherent heart surrenders gastrointestinal abnormalities [5-7]. Children with Down syndrome are at higher risk for visual and hearing impairments, thyroid problems, and leukemia, and may have a loss of hearing due to fluid in the middle ear, a nerve defect, or both. The mental issue associated with Down's syndrome is mental retardation ranging from mild to moderate, and moderate to severe. There is no way to predict the mental development of a child with Down's syndrome based on physical appearance.

#### Causes and risk factors

For many people, each cell in the body has 23 sets of chromosomes. In each pair, one chromosome comes from the

female and the other from the male. However, with Down's syndrome, something turns out badly and you get an additional duplicate of chromosome 21. That means you have three copies of two, which leads to the symptoms of Down syndrome. Doctors aren't sure why this happens. There's no connection to anything in the climate or anything the parents did or didn't do. While specialists don't have the foggiest idea of what causes it, they truly do realize that, women 35 or more aged have a higher possibility of having a child with Down's syndrome. If you have already had a child with Down syndrome, you're more likely to have another one who has it as well. It's rare. However, it is feasible to pass Down syndrome from parent to youngster. Sometimes a parent has what specialists call "translocated" genes [8]. That implies a portion of genes aren't in their regular spot, maybe on an alternate chromosome from where they would for the most part be found. The parents don't have Down syndrome because they have the right number of genes, but their child may have what's called "Translocation Down Syndrome". Not everyone with Translocation Down Syndrome gets it from their parents-it might likewise occur by some coincidence [9,10].

#### In the first trimester, it includes:

Blood tests: Doctor will gauge the levels of protein called PAPP-A and a hormone called hCG in blood. Anything out of the typical reach could mean an issue with the child. Assuming you're at high danger for Down syndrome, they may likewise search for DNA from the child in blood, which they can look at for chromosome irregularities.

**Ultrasound:** Primary care physician will check out an image of child and measure the folds of tissue at the rare of their neck. Babies with Down syndrome tend to have extra fluid here.

### In the second trimester, you may have:

Blood tests: Either a triple or quad-screen test estimates different substances in blood, including the protein AFP and the chemical estriol. Those levels joined with the outcomes from the main trimester tests, provide primary care physician with a decent gauge of the possibilities child has Down syndrome.

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**Ultrasound:** When baby is more developed, enhanced ultrasound can show some of the physical features of Down syndrome. Different sorts of tests can analyze Down syndrome before child is brought into the world by really taking a look at an example of their DNA for an additional chromosome 21.

# CONCLUSION

There are the best rehabilitation centers in UAE that help in dealing with children with these issues. In most cases, specialists diagnose Down syndrome even before the birth of the child giving parents legitimate data and information in regards to the matter. Sometimes the syndrome is so mild that it will go unnoticed, whereas certain other cases require good therapies.

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