

Timely genetic screening crucial for Down's Syndrome in babies

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On World Down's Syndrome Day, awareness must be raised on the non-invasive and effective screening tests available



According to recent statistics, the incidence of Down's Syndrome in India is 1 per 850- 900 live births. It is one of the most commonly occurring chromosomal abnormalities in India and children born with the condition have an average life expectancy of 50 to 60 years.

Down's Syndrome or Trisomy 21 is a genetic condition marked by an extra copy of chromosome 21. It is associated with intellectual disability, a characteristic facial appearance, and a weak muscle tone (hypotonia). While there is no exact reason for its occurrence, the age of the mother at the time of conception and family history of the condition may be two of the risk factors.

Speaking about this, Dheeraj Jain, Founder, Redcliffe Lifesciences, said, "Genetic disorders such as Down's Syndrome are a very serious health burden and there is a need to work towards making available informed choices to parents as early as 10th week of the pregnancy. With advent of Next Generation Sequencing and increasing awareness, there are non-invasive screenings such as DNA based Prenatal Genetic Testing available today that can help identifying three types of trisomies one of which is Trisomy 21, the anomaly behind Down's Syndrome. At Redcliffe, we offer genetic screening options at our in-house lab to enable both clinicians and parents to understand the risk of their child being born with the condition. An NGS routine genetic testing is comprehensive and can detect all types of genetic mutations. At Redcliffe we have made it affordable to masses with fastest turnaround time to the clinicians, in order to break the myth that samples sent to western countries are very expensive test"

Some signs in a child with this condition include low muscle tone, enlarged tongue, and a flat facial profile. They also experience intellectual disability apart from health issues such as respiratory infections, heart defects and hearing problems.

Adding further, Padma Shri Awardee, Dr K K Aggarwal, President, Heart Care Foundation of India (HCFI) and Confederation of Medical Associations of Asia and Oceania (CMAAO), said, "The space of genetic testing is growing at a rapid pace and more intensive research is being carried out. It is important to raise awareness on how beneficial these tests are when it comes to detecting abnormalities such as Down's Syndrome early. Making screening compulsory at a nation-wide level can

be the first step in this regard.”

Although it may not be possible to prevent all birth defects, there are steps women can take to reduce the likelihood. Some of these include undertaking regular check-ups, routine exercises, consuming folic acid daily before and during pregnancy, eating a healthy diet, and avoiding habits like smoking and drinking.