

The lure for perfect baby

Arun (name changed), a 32-year-old male architect, presented with history of short fingers and toes since birth and impaired fist formation due to stiffness of the finger joints. The family history revealed that his mother and younger sister Aarti (name changed) were similarly affected with mild variations in the clinical features. Genetic testing at DBT's Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad, revealed a new mutation in *NOG* gene (p.Arg204Gln) known to cause a syndrome of skeletal abnormalities caused by fusion of the bones in the wrist, ankles, fingers and toes known as Tarsal-carpal coalition syndrome (TCC).



The TCC is inherited in autosomal dominant fashion and risk of transmission to progeny of affected individual is 50%. For many individuals with TCC, symptoms are relieved with simple treatments of physical therapy and ultimately they learn to function with the disability. For patients with severe symptoms that do not respond to simple treatments and if the disability interferes with their daily activities, surgery may be recommended. Arun was fine with his abnormality and leading a normal life. Once the mutation is identified, families have the option to look for the presence of the disease causing mutation in the next child through genetic testing before the birth of the child and plan accordingly.

Aarti wanted to go for prenatal diagnosis and abort in case the baby had the same problem although she had herself lived a normal life with the same problem. This report brings out the dilemma faced by the geneticist in counseling such families. Although many abnormalities like cleft lip/palate, TCC etc can be easily treated with simple surgery after birth of child, the couple may still opt for prenatal diagnosis and termination of pregnancy due to social pressures of

having a perfect baby. The question is, whether it is justified to do prenatal diagnosis and termination of pregnancy for diseases where symptoms are manageable and an individual can live all his/her life with minimal burden.

Human body is made up of trillions of cells. In each cell, there are 46 chromosomes, found in 23 matching pairs. Half of the chromosomes are inherited from an individual's mother and half from their father. These chromosomes carry our DNA, or genes, which are the instructions for how we look and how our body develops and functions. When a harmful change occurs in these instructions (mutation or variation), it can change the way a baby develops. Babies with genetic disorders can be at risk of slow mental and physical development, physical abnormalities and lifelong illnesses. However, every genetic disease need not cause significant problems or show severe symptoms but still people want perfect babies. The work was published in *American Journal of Medical Genetics*.

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