MDCRC - World Duchenne Awareness Day

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DNA testing suggested for muscular dystrophy diagnosis

DC CORRESPONDENT COIMBATORE, SEPT. 8

As many laboratories in India still practise the painful muscle biopsy procedure to diagnose muscular dystrophies or Duchenne musclar dystrophy (DMD) common among children, boys and young men, the molecular diagnostics, counselling, care & research centre (MDCCRC), that functions in a city hospital, observed that a non-invasive, three-level DNA testing would be ideal for diagnosis.

Functioning out of Kurunji Hospital, the MDC-CRC. recognised by the department of health and family welfare. Government of Tamil Nadu, offers services like molecular diagnosis, counselling and care, all offered free of cost to patients, under one roof.

According to the founder and head of MDCCRC, Dr B.R. Lakshmi, prevention of this disorder plays a crucial role in the victim as DMD is a life threatening, genetic, neuromuscular disorder affecting boys. Duchenne patients progressively lose their physical functions before they inevitably

Non-invasive, threelevel DNA testing is an ideal alternative to the painful muscle biopsy procedure to diagnose muscular dystrophies

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die. They suffer respiratory problems or heart failure and most of them do no reach adulthood at all. Duchenne muscular dystrophy is one of the most common fatal genetic disorders which affects, on an average, one in every 3,500 newborn boys, with very few girls suffering from the disease, according to WHO.

"The non-invasive DNA testing involves three levels and when results from the first level are inconclusive, testing at the second level is required and then the third. Based on the results of molecular diagnosis, genetic counseling is offered wherein the family is enabled to understand the disorder, etiology of the disease, genetic inheritance pattern, risk factors, importance of carrier diagnosis, guidance towards prenatal diagnosis and options available for the management of the disorder."

Steroids are the only options to manage the disorder. The effect, however, varies from child to child but there are some studies revealing that some boys are able to walk for a longer duration with the help of steroids.

Comprehensive lines for the management of DMD were published by a consortium of leading neuromuscular specialists across the world in 2010. Based on these international guidelines, MDCCRC offers a comprehensive, multi-disciplinary clinical care programme involving pediatrics, neurology, orthopedics, physiatry, pulmonology, cardiology and psychosocial management for DMDaffected children.

As the disorder does not have a cure till date, MDC-CRC has embarked on a collaboration with the Arya Vaidya Pharmacy (AVP), to understand the therapeutic advantage that Ayurveda can offer. MDCCRC's effort with the National Rural Health Mission has established the fact that the prevalence of DMD is a stunning 2.4 times more than that of global figures.