

A Mother's Intuition, a Doctor's Insight: Decades-Long Quest Ends with Genetic Diagnosis

This case study revolves around a family consisting of a couple and their three adult sons, aged 31, 32, and 33 years. The sons exhibited developmental delays, intellectual disabilities, and other health issues since childhood. Despite multiple visits to various doctors over a span of 30 years, the cause of their condition remained unknown. Eventually, a crucial genetic diagnosis shed light on their condition, leading to a better understanding and potential options for the family's future.

The family's three sons exhibited common traits such as developmental delays, intellectual disabilities, hyperactivity, and obesity. Despite seemingly normal antenatal and post-natal histories, their early life was marked by recurrent upper respiratory infections, delayed milestones, and neurological abnormalities. The family's persistence in seeking medical advice and undergoing tests demonstrated their unwavering commitment to understanding their sons' condition.

Over time, the family's hopes diminished as they faced countless doctor visits and even stem cell transplantation, a desperate attempt to find a solution. The turning point came when they sought medical consultation with the author, who, based on their comprehensive history and symptoms, suspected a genetic issue and recommended **appropriate** genetic testing.

Genetic testing revealed that all three affected sons and their mother carried a CUL4B mutation. This X-linked recessive condition explained the siblings' shared symptoms and developmental challenges. The diagnosis not only provided clarity about the condition but also offered insights into potential future outcomes and management strategies.

The family received detailed genetic counseling to understand the implications of the diagnosis. They learned that the CUL4B mutation was responsible for their sons' conditions and that their mother was a carrier of the mutation. This newfound knowledge eased their anxieties and provided a clearer understanding of the family's genetic makeup.

The family expressed gratitude for the diagnosis that brought clarity to their decades-long struggle. With knowledge about the genetic cause, the family could finally move forward with a sense of direction. The parents felt relieved that future pregnancies could be planned with confidence, knowing the genetic basis of the condition and the risk factors involved.

The case study underscores the importance of pursuing a definitive diagnosis, especially when faced with complex and longstanding medical issues. It highlights the value of genetic testing in identifying underlying genetic mutations responsible for various health conditions. The case also serves as a reminder that accurate diagnosis can lead to informed decisions, better management strategies, and more hopeful outlooks for affected families.

Talking about this case, **Dr. Sheetal Goyal, Brain And Nerve care, Wockhardt Hospitals, Mumbai Central**, said "This case made me witness personally the transformative power of accurate genetic diagnosis. It not only provides answers to long-standing medical mysteries but also offers a guiding light for families seeking understanding and direction. Through the lens of genetics, we unveil the intricate stories written within our genes, paving the way for personalized care and empowering families to navigate their journey with knowledge and hope."

The journey of this family showcases the significance of persistence in seeking answers and the pivotal role of genetic diagnosis in unraveling complex medical conditions. It emphasizes the importance of medical professionals' expertise in recognizing patterns, asking the right questions, and recommending appropriate diagnostic tests. Ultimately, the timely genetic diagnosis not only brought relief to the family but also illuminated a path forward, allowing them to make informed decisions about their future.