

## Genetic Diseases Work Up Pathways

**Kuldeep Singh**

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Though genetic disorders are individually rare but assume a heavy burden on society in terms of morbidity, mortality and psychosocial impact. Many of these disorders present at birth as Congenital Anomalies, also known as birth defects, which are structural or functional abnormalities, including metabolic disorders. They are a diverse group of disorders of prenatal origin, which can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens or micronutrient malnutrition. Many of these disorders are so rare that physician or health professional may be managing for the first time. It requires consulting many resources, colleagues and experts to arrive at a diagnosis. According to World Health Organization (WHO) report in 2010, it is estimated that 2,70,000 deaths are attributable to congenital anomalies[1]. The common eight emotional effects of genetic diseases were identified by McAllister *et al* [2] as: anxiety, worry about risks to children, guilt, anger, uncertainty, sadness and grief, depression, and redemptive adjustment. The problem of genetic conditions has been highlighted by many workers in India also[3, 4, 5]. Clinical diagnosis still remains the cornerstone for optimal management of the disorders. A clinical contact not only helps in building rapport with the family but also helps in understanding the psychological makeup and ability to respond to stressful information.

Apart from clinical suspicion, arrays of diagnostic services are available for confirming genetic conditions. Some of these services are extremely cost prohibitive. Bridging the gap between clinical services and genetic data is possible. The clinicians can rely on cascade testing using algorithmic approach to minimize the cost to the family. Many examples exist worldwide which have shown

benefits. The UK strategy focus on empowering those affected, identifying and preventing rare conditions, early intervention, coordination of care and research [6]. Globally organizations are joining hands to help children with rare diseases. Last day of February is celebrated as a Rare Diseases day. Overall the cost evidence of rare disorders appears to be scarce[7].

In this issue of the journal, we have articles related to finding genetic damage through Comet assays to focus on some common conditions like Asthma and Thalassemia. The issue also highlights a rare ophthalmic conditions and subtle presentations of common cytogenetic disorder.

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