GENETICS

CONSTITUTIONAL CHROMOSOMES – FROM KARYOTYPING TO CHROMOSOME MICROARRAY IN PEDIATRIC PRACTICE

***Siddaramappa J Patil**
****Pooja N Rao**

Abstract: *Chromosomal abnormalities are one of the causes of multiple malformations with or without intellectual disability in children. Knowing and identifying underlying chromosomal abnormality requires prior knowledge of clinical chromosomal phenotype (indications to study chromosomes) and selection of appropriate chromosomal tests to diagnose. Definitive diagnosis forms the basis for proper management and genetic counselling. This article discusses briefly about the types of constitutional chromosomal abnormalities, methods to study chromosomes and management including genetic counselling.*

Keywords: *Karyotype, Chromosome analysis, Chromosome microarray, Congenital anomalies, Intellectual disability, Molecular cytogenetics.*

* Consultant - Clinical Genetics

** Research Associate, Cytogenetics Laboratory,
Divison of Medical Genetics,
Mazumdar Shaw Medical Center,
Narayana Health City,
Bangalore.

email: drsjpatil@gmail.com

Points to Remember

- ***Common clinical presentation of chromosomal disorders in children includes congenital malformations, intellectual disability, growth abnormalities and facial dysmorphism.***
- ***Facial dysmorphism forms an important clue to diagnose and investigate for chromosomal disorder.***
- ***Resolution varies among the various chromosomal tests available to diagnose chromosomal disorders. Knowledge on use and limitation of each chromosomal test helps in selecting the appropriate test.***

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