

Towards a future of proactive, personalized pediatric care

DGx **Pediatric Comprehensive**

Using NGS



What is DGx Pediatric Comprehensive test?

A genetic screening test performed to identify inherited / genetic disorder in newborns and children.

- Identifies genetic, metabolic, hormonal, and functional disorders
- Facilitates timely intervention to prevent severe health complications / death

Why is DGx Pediatric Comprehensive test needed?



4 million children are born with some genetic abnormality



Increased association of infant mortality with genetic disorders
(owen et al)¹



In India, **5-15%** of the sick newborn have a metabolic problem
(Manju et al)²



Sudden Infant Death Syndrome (SIDS) due to metabolic disorders
CAN BE TREATED
(Van Rijt et al)³

DGx Pediatric Comprehensive test: Accelerating screening diagnosis and treatment

Identify Earlier

- Shortening the diagnostic odyssey
- Cost effective, shorter turn-around times

Intervene Earlier

- Prevent associated avoidable morbidity and mortality
- Intervene to change the course of life from birth

Access Earlier

- Access to precision medicine
- Clinically actionable reports for 200+ disorders and 450+ genes

Who should get tested?

DGx Pediatric Comprehensive test is recommended for **all newborns and children**, regardless of their perceived health or risk factors.

Essential for:

**ALL
Newborns**

Especially for:



Premature babies



Babies with low birth-weight



Babies / children with a family history of genetic disorders



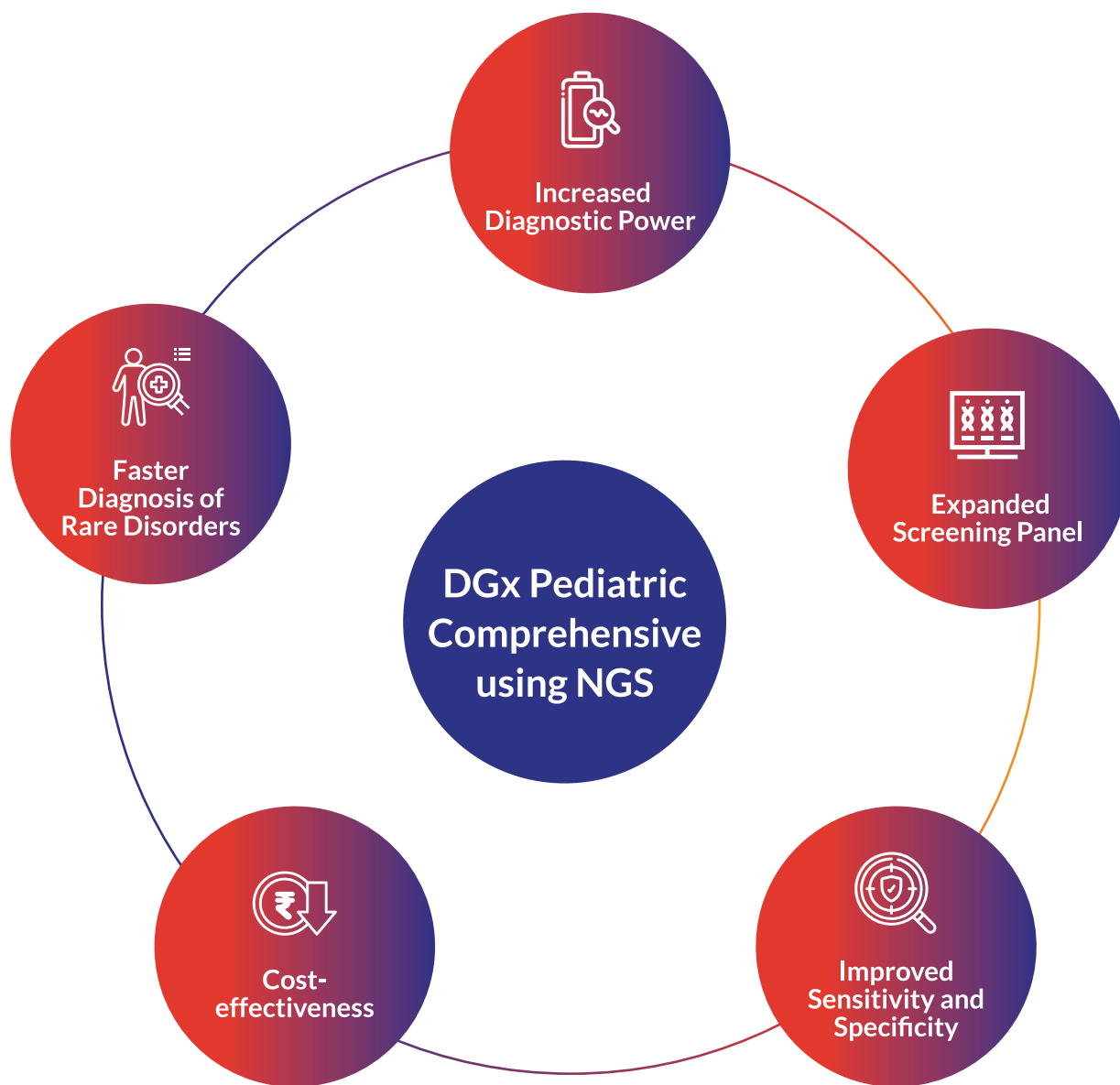
Children (0-18 years old)

Examples of conditions screened

- **Metabolic Disorders:** If untreated, can lead to intellectual disabilities (E.g., Phenylketonuria (PKU), Galactosemia, Homocystinuria, Maple Syrup Urine Disease)
- **Endocrine Disorders:** Can result in growth delays but is treatable with medication (E.g., Congenital Hypothyroidism, Congenital adrenal hyperplasia (CAH))
- **Haemoglobinopathies:** Ensuring early management to prevent crises (E.g., Sickle cell anemia, Beta-thalassemia)
- **Immunodeficiencies:** Facilitating timely intervention to cure the condition (E.g., Severe Combined Immunodeficiency (SCID))
- **Cystic Fibrosis:** Providing early treatment to prevent complications and improve survival rates
- **Lysosomal Storage Disorders:** Rare conditions (E.g., Pompe disease, Mucopolysaccharidosis type I (MPS I), Gaucher disease)
- **Fatty Acid Oxidation Disorders:** Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- **Amino Acid Disorders:** Detect issues with the breakdown of amino acids (E.g., Tyrosinemia, Argininosuccinic aciduria)
- **Organic Acid Disorders:** Identify disruptions in organic acid metabolism (E.g., Isovaleric acidemia (IVA), Methylmalonic acidemia (MMA))
- **Neurological Disorder:** Ensuring early detection of degeneration of motor neurons (E.g., Spinal Muscular Atrophy (SMA))



Revolutionizing genetic disorder diagnostics in pediatric population



References:

1. Owen et al. JAMA Network Open(2023)6(2):e2254069.
2. Manju et al. BMJ Public Health (2024);2:e001459.
3. Van Rijt et al. Neonatology (2016) 109 (4): 297–302.



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