Towards a future of proactive, personalized pediatric care

DGx
Pediatric Screening

Using NGS

What is DGx Pediatric Screening 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 Screening test needed?

4 million children are born with some genetic abnormality

Increased association of infant mortality with gentic 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 Screening 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 Screening test is recommended for **all newborns and children,** regardless of their perceived health or risk factors.

Essential for:

ALL
Newborns

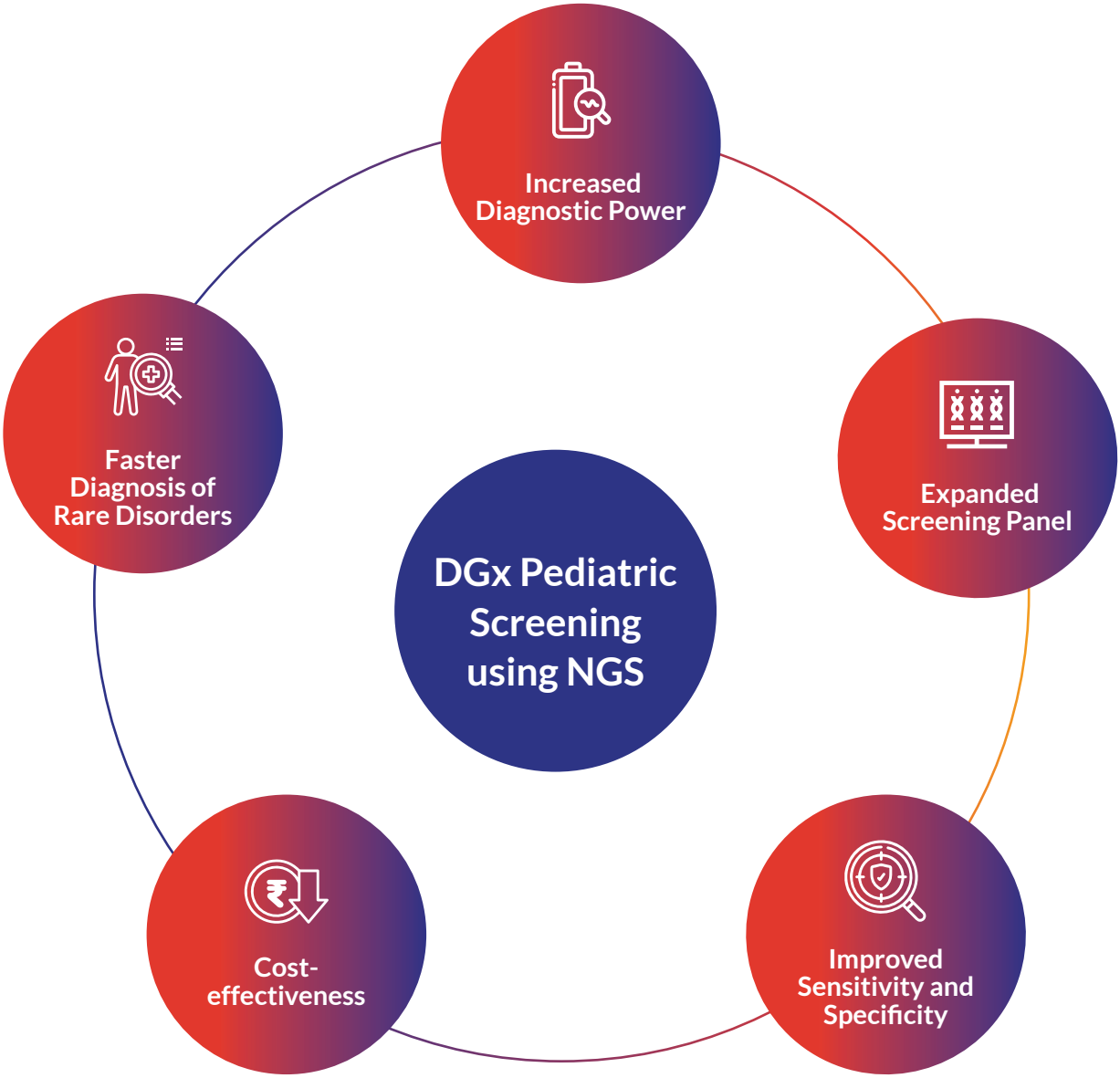
Especially for:

- 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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