THE ROLE OF NEWBORN SCREENING IN RARE DISEASE DETECTION

Advancing Early Diagnosis & Treatment for Better Outcomes

INTRODUCTION

What is Newborn Screening (NBS)?

- A public health program that tests newborns for genetic, metabolic, and congenital disorders

Why is it Important?

- Enables early diagnosis and intervention
- Prevents severe disability, organ damage, or death

HOW NEWBORN SCREENING WORKS

- Process Overview:
  a. Blood Sample Collection – Heel prick within 24–48 hours after birth
  b. Laboratory Testing – Screens for multiple conditions
  c. Follow-up & Diagnosis – Further tests if results are abnormal
  d. Early Treatment Initiation – Prevents complications

IMPACT OF NEWBORN SCREENING

01 BETTER QUALITY OF LIFE

02 IMPROVED SURVIVAL RATES

03 REDUCED HEALTHCARE COSTS

FUTURE OF NEWBORN SCREENING

ADVANCES IN GENOMICS & AI

- Whole genome sequencing

EXPANSION OF SCREENING PANELS

- More rare diseases to be included

GLOBAL COLLABORATION

- Standardized policies for better implementation

DID YOU KNOW

Rare disease organizations like IndoUSrare, NORD, and Global Gene celebrate the vital role of early detection in giving every newborn the healthiest start. Screening for rare genetic and metabolic conditions can provide timely interventions that pave the way for a brighter, healthier future.