# A MANAGEABLE CONDITION IS IDENTIFIED IN APPROXIMATELY 1 in 300 NEWBORNS\*

Oreana neonatal screening can detect **treatable** or **manageable** conditions **before the appearance of symptoms**. With early treatment and management, symptoms can be averted, and health problems can be minimized or prevented.

Oreana can also be used for newborns, infants and children who show symptoms of a genetic disease which could be difficult to identify. In these cases, Oreana can lead to **the appropriate therapy** that will **benefit** the affected child.

## WHY PERFORM **Oreana** IN ADDITION TO NATIONAL NEWBORN SCREENING?

National newborn screening programs offer great advantages to infants, their families and national health systems. However, since they are subjected to each country's regulations, the structure of the healthcare system, and available funds, there are vast differences in the number of conditions each country tests for and in the technology they use. As a result, countries throughout the world perform newborn screening ranging from only 2 disorders to over 35.

Oreana neonatal screening tests for 106 genetic disorders that are either treatable or can benefit from early detection. Among others, Oreana tests for metabolic, endocrine, haemoglobin and hearing loss disorders, whose early detection and management can lead to therapeutic treatments that could reduce the severity of symptoms and improve the prognosis and life quality of the affected infant.

\*Data collected from Centers for Disease Control and Prevention (2012); CDC Grand Rounds: Newborn screening and improved outcomes, and is based on screening of 29 diseases included in the core conditions of the Recommended Uniform Screening Panel.

### HOW IS **Oreana** NEONATAL SCREENING PERFORMED?



ASK YOUR PEDIATRICIAN ABOUT

Oreana



YOUR DOCTOR WILL COLLECT THE SAMPLE FROM YOUR BABY USING A BUCCAL SWAB



THE SAMPLE WILL BE SENT TO NIPD Genetics



THE SAMPLE WILL BE ANALYZED IN OUR LABORATORY



RESULTS WILL BE SENT TO YOUR DOCTOR WITHIN 2-3 WEEKS OF SAMPLE RECEIPT

#### MORF QUESTIONS?

If you have additional questions or concerns, please ask your doctor. You can also contact us:



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### WHAT IS **Oreana** NEONATAL SCREENING?

Oreana is a genetic test that screens for genetic conditions which affect normal development in newborns, infants or young children. Oreana is a beneficial test for:

- Asymptomatic infants, for early identification of conditions which may not cause any symptoms initially, and babies may not show any signs of illness. However, once symptoms begin, their damage may be irreparable and the health of the infant can deteriorate quickly, so early detection is beneficial.
- Symptomatic infants which have signs or symptoms of disease that can be difficult to be clearly identified, due to complexity or variability of symptoms. In these cases, Oreana could correctly identify the genetic mutation (change in the DNA) responsible for the disease, and can lead to the affected child receiving the optimal treatment.

Most of the disorders included in Oreana are either **treatable** or have a **simple management plan** such as following dietary restrictions and taking prophylactic measures. When initiated early, symptoms can be **prevented** or their severity may be reduced, improving the prognosis and life expectancy of affected infants.

### WHAT ARE THE BENEFITS OF Oreana NEONATAL SCREENING?

With Oreana, early detection of life-altering disorders can benefit the infant's health and quality of life. The initial symptoms for many of these disorders include vomiting, lack of energy, breathing problems, feeding difficulties, and episodes of metabolic crises that require urgent care. Although treatment can begin when infants first start showing symptoms, which may be a few weeks or months after birth, their growth and development will already be impacted. Early detection and starting treatment as soon as possible is the most favorable way to prevent symptoms or minimize the impact of the condition.

Early detection can

- ◆ Prevent or minimize symptoms
- Reduce the impact of the condition
- Reduce the time spent for diagnosis, and prevent a late diagnosis
- Permit early clinical interventions, treatment and management, which will be more beneficial for the infant
- ◆ Enable potential participation of the infant in experimental therapies or clinical trials



Oreana tests for **106 genetic conditions**, whose **early detection** can prevent or reduce serious consequences, such as developmental delay, cognitive impairment, neurological and physical problems, and premature death. The conditions tested may be inherited to children from carrier parents who are unaware of their carrier status, with or without a prior family history of that disorder.



#### **METABOLIC DISORDERS**

Lifelong dietary management is required. Early diagnosis and interventions can help prevent symptoms and improve prognosis.



#### **ENDOCRINE DISORDERS**

Early medical treatment and interventions can improve thyroid function, growth, development and metabolism. Lifelong treatment and follow-up by specialists is necessary.



#### **HAEMOGLOBIN DISORDERS**

Medical treatment, dietary management and transfusions are critical and needed lifelong.



#### **HEARING LOSS DISORDERS**

Hearing aids, cochlear implants or other services can help with creating the best management plan. Early interventions can improve the speech, language and social skills of affected children.



### IMMUNODEFICIENCY, PULMONARY OR MUSCULOSKELETAL DISORDERS

Including Cystic Fibrosis, Severe Combined Immunodeficiencies and Spinal Muscular Atrophy. Early detection is critical for prompt medical treatment, such as bone marrow transplantation, and better prognosis.

Oreana tests for 142 genes and 106 Autosomal Recessive, Autosomal Dominant and X-Linked disorders. The disorders tested by Oreana are included in the 'Recommended Uniform Screening Panel', a list of disorders selected based upon the recommendations of the American College of Medical Genetics (ACMG) for newborn screening. Additional clinically significant, manageable disorders were added to the Oreana panel.

### WHY CHOOSE **Oreana** NEONATAL SCREENING?



Results are reported quickly, so if necessary, your baby can start beneficial treatment as soon as possible.



Tests for disease-causing genetic mutations that cause serious health problems.



Superior technology that yields accurate results.



Non-invasive and painless for the infant.



Complements newborn screening programs that test a smaller number of disorders.

#### **HOW IS TESTING PERFORMED?**

Testing is **quick and safe** for your baby. A healthcare practitioner will use a specialized, soft swab to collect sample from the inside of the cheek of the infant, by moving it around in circular movements. The procedure is painless and is completed in a few seconds. The results will be reported back to your healthcare practitioner.

#### WHEN IS THE IDEAL TIME FOR TESTING?

Your baby can get tested **anytime after birth**, up until early childhood. Ideally, the earlier the infant is tested, the sooner you will know whether they are affected, and start **beneficial treatments** that can prevent the onset of symptoms or minimize the impact of the disease.