Grants and Publications

Several parts of this study are funded by grants from National Institutes of Health and the March of Dimes Foundation. This study also is supported by the Children's Mercy Research Institute.

Some of the important publications that have arisen from our work showing a possible role of genetic risk factors in neonatal disease are summarized below.

1. SIGIRR Mutations in Premature Infants with Necrotizing Enterocolitis. Dr. Sampath's data provided new insight into the probable cause of NEC and support the theory that inherited defects in genes that stop intestinal innate immune signaling can contribute to NEC.
2. A Functional ATG16L1 (T300A) Variant is Associated with Necrotizing Enterocolitis in Premature Infants. Dr. Sampath's study reports a new link between a hypo-morphic variant in an autophagy gene (ATG16L1) and NEC in premature infants. The data suggests that decreased autophagy arising from genetic variants may give protection against NEC.
3. Rare Genetic Variants in Immune Genes and Neonatal Herpes Simplex Viral Infections. This study is the first to report that newborn babies with life-threatening herpes viral infections might have defects in immune genes that help fight viral infections.
4. Anti-Oxidant Response Genes Sequence Variants and BPD Susceptibility in VLBW Infants. This large study did not demonstrate an association between genetic variants in genes that regulate response to oxygen toxicity and vulnerability to BPD in premature infants.

HOW DO YOU FIND OUT MORE?

Contact our team by email or phone!

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HOW YOU CAN HELP US STUDY THE GENETIC LINK TO NEWBORN DISEASES

Why study genetic risk factors?

More and more often, doctors and researchers are understanding that there may be a genetic reason why some infants have a greater risk of becoming sick from certain diseases. Some of these diseases are directly or indirectly caused by germs, including bacteria, viruses or fungi. If we can identify the genetic links that make some infants more likely to become sick from these germs, we may be able to target treatment to these higher-risk babies in the future, which could prevent severe illness and even death.

Why is genetic research important and what does it involve?

Venkatesh Sampath, MD, is leading the research team handling the Genetic Susceptibility to Neonatal Diseases study. This team is searching for the genetic risk factors that might make some babies more likely than others to develop certain neonatal diseases.

In premature infants, the main diseases that germs cause are:

- Neonatal sepsis, a blood infection in an infant less than 90 days old.
- Necrotizing enterocolitis (NEC), damage to the intestinal tract causing the death of intestinal tissue.
- Bronchopulmonary dysplasia (BPD), severe lung disease in premature infants.

In full-term infants, bacteria such as Group B streptococcus and viruses such as herpes simplex can cause serious infections.

These diseases are major causes of illness and death in babies. Early studies suggest that genetic differences in the immune system can affect how likely an infant is to get these diseases, and how well they respond to treatment. (See Grants and Publications).

Our team also is looking for genetic risk factors that can cause babies to have rare, severe lung or other organ disease. These diseases often require life support and long hospital stays.

Finding the genetic risk factors could make it possible for us to screen babies for the risk of several diseases, and potentially prevent or treat them before they become life-threatening.

What does this mean for your child?

The study team is looking for genetic differences that might make certain babies more likely to become sick from NEC, BPD and specific blood infections during the birth to 12-month-old period.

We are approaching you because your child either has one of these diseases, is at risk of developing one of these diseases, or is a healthy infant with no suspected genetic risk factors.

How can your child help?

When you give approval for your infant to participate in this research study, the team collects a small amount (0.5ml) of blood from your baby. This sample is only collected once. It is then de-identified. This means that any information which could connect your child to their study sample is removed and replaced with non-traceable serial numbers to protect you and your child's identity.

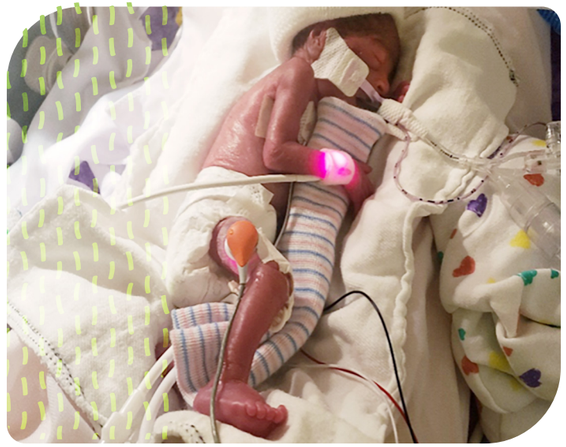
There is no direct benefit to your child and little risk related to helping with the study, but the impact for babies in the future could be great!

Your Research Team

A committed group is required to successfully conduct a study of this size. We are close to recruiting 1000 infants from sites across the United States and the United Kingdom. We hope to recruit 2000 in a few years. Our team is led by Venkatesh Sampath, MD, Principal Investigator, Neonatology (high-risk baby doctor) and research scientist. Dr. Sampath has a special interest in understanding genetic and other risk factors that cause newborns to have these diseases.

Other members of the research team include:

- Dr. Venkatesh Sampath, MD, Neonatologist, Physician Scientist
- Dr. Lovya George, MD, Neonatologist
- Dr. Gangaram Akangire MD, MS, Neonatologist
- Heather Menden, MS, Research Associate Masters, Lab Manager
- Staci Elliott, RN, MSN, NNP-BC, Clinical Research Coordinator, Neonatal Nurse Practitioner
- Anne Holmes, RN, MSN, MBA-HCA, CCRC, Clinical Research Coordinator
- Miah Ruffin, Clinical Trials Coordinator
- Laboratory scientists and other collaborators including the Genome Center at Children's Mercy Research Institute



A Parent's Perspective on Research

London Edmondson was born at 25 weeks and weighed less than 1 pound. She is a research participant in Dr. Sampath's study at Children's Mercy Kansas City.

"We are so thankful to assist future premature babies and we know our contributions to this research study will improve their quality of life and further advancements in medical research."
– Charlette Edmondson, London's mom

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