MEDICAL BREAKTHROUGHS
RESEARCH SUMMARY

TOPIC: INFILTR8: RARE DISEASE ATTACKS A CHILD'S IMMUNE SYSTEM
REPORT: **MB #4986**

BACKGROUND: Toll-like receptors (TLR) sense distinct pathogen associated molecular patterns (PAMPs) and initiate inflammatory reactions important for natural and adaptive defense. Human TLR8 is defined as a functional cleavage product in endosomes of monocytes and macrophages. The RNA degradation products uridine and short oligomers bind cooperatively at two distinct sites in the N-terminal domain. This induces a conformational change of the pre-formed TLR8-dimer leading to MyD88 recruitment and signaling. (Source: https://www.frontiersin.org/articles/10.3389/fimmu.2019.01209/full)

DIAGNOSING IMMUNE SYSTEM ISSUES IN CHILDREN: One of the most common signs of INFILTR8 is having infections that are more frequent, longer lasting or harder to treat than are the infections of someone with a normal immune system. Signs and symptoms can vary from person to person and can include frequent and recurrent pneumonia, bronchitis, sinus infections, ear infections, meningitis or skin infections. Signs can also include Inflammation and infection of internal organs, blood disorders, digestive problems, and delayed growth and development. Doctors may run blood tests to determine if there are normal levels of infection-fighting proteins in your blood and measure the levels of blood cells and immune system cells. Parents might want to be tested for certain immunodeficiency disorders during future pregnancies if they have a child that has experienced issues with their immune system. (Source: https://www.mayoclinic.org/diseases-conditions/primary-immunodeficiency/symptoms-causes/syc-20376905)

NEW STUDY: Megan A. Cooper, MD, PhD, an associate professor of pediatrics at Washington University School of Medicine in St. Louis and a postdoctoral fellow in her laboratory, Jahnavi Aluri, PhD recently identified an additional child with this rare disease and are actively searching for more. More than 30 scientists from a dozen research and medical centers contributed to the TLR8 study. The disorder was named by the fifth patient enrolled in the study, Evan Brandon of Tuscaloosa, Ala. In the study, all six patients with TLR8 variations also suffered from bone marrow failure. Marrow, a soft, spongy tissue found in the center of bones, produces red blood cells, white blood cells and platelets.
(Source: https://medicine.wustl.edu/news/scientists-discover-rare-genetic-condition-that-attacks-kids-immune-systems/)

FOR MORE INFORMATION ON THIS REPORT, PLEASE CONTACT:

DIANE DUKE WILLIAMS
williamdia@wustl.edu

If this story or any other Ivanhoe story has impacted your life or prompted you or someone you know to seek or change treatments, please let us know by contacting Marjorie Bekaert Thomas at mthomas@ivanhoe.com