

Gene found that raises risk of childhood ear infections

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Researchers have discovered a gene region that raises the risk a child will have a middle ear infection, known to doctors as acute otitis media (AOM)—and known to parents as one reason for a screaming, unhappy



preschooler. The finding may offer an early clue to helping doctors develop more effective treatments to prevent one of the most common childhood illnesses.

"Parents and pediatricians are all too familiar with this painful childhood ear infection—it's the most frequent reason children receive antibiotics," said study leader Hakon Hakonarson, MD, PhD, director of the Center for Applied Genomics at The Children's Hospital of Philadelphia (CHOP). "Although microbes cause this condition, it's been well known that genetics also plays a role. This is the first and largest genetic study focused on risk susceptibility for acute otitis media."

Hakonarson and colleagues from CHOP collaborated with Dutch researchers led by Gijs van Ingen and colleagues from the University Medical Center, Rotterdam, in the study published online Sept. 28 in *Nature Communications*.

The researchers performed a genome-wide association study (GWAS) on two discovery cohorts with DNA samples from 11,000 children. They found that an association between AOM and a site on chromosome 6 containing the gene *FNDC1*, and then replicated the finding in an independent pediatric cohort with data from 2000 children. Co-first author Jin Li, PhD, of CHOP, was the lead analyst on the study.

In further studies, the scientists showed that the mouse gene corresponding to *FNDC1* was expressed in the animal's middle ear. "Although the gene's function in humans has not been well studied, we do know that *FNDC1* codes for a protein with a role in inflammation," said Hakonarson.

Hakonarson added that learning more about the biological interactions between genes and pathogens involved in AOM, researchers hope to develop more specific therapies for the childhood infection, while



learning which children are most susceptible to the disease. "As with many other diseases, early medical intervention may offer the greatest benefits," he said.

More information: Gijs van Ingen et al, Genome-wide association study for acute otitis media in children identifies FNDC1 as disease contributing gene, *Nature Communications* (2016). DOI: 10.1038/ncomms12792

Provided by Children's Hospital of Philadelphia

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