

New research shows strong heredity component to middle ear cholesteatoma

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Atticuscholesteatoma. Credit: Karolinska Institutet

A nationwide Swedish study including more than 10,000 cases of cholesteatoma, a lesion in the middle ear, shows a strong hereditary component to the disease.

Cholesteatoma is a benign growing retraction pocket in the [middle ear](#) originating from the [tympanic membrane](#). During growth, the surrounding tissue such as ossicles, surrounding [bone](#) and nerves are affected, with the risk of damaging the bone toward the inner ear or brain with hearing loss and risk of intracranial infections as a result. Every year, 600–700 individuals are treated with surgical removal of the disease in Sweden. However, residual problems with hearing loss and taste disturbances after surgery can accrue.

The current study is a case-controlled study including more than 10,000 individuals who underwent primary surgery for cholesteatoma over a 30-year period. The results show an almost fourfold higher risk of cholesteatoma in individuals with a first-degree relative surgically treated for the disease. It is a small percentage of the total number of individuals who have any [heredity](#) connection, but the group can be an important source of further knowledge about the [disease](#). The research group, SwedEar, is now continuing with more studies on clinical and genetic aspects of hereditary cholesteatoma.

The study is the first of its kind to investigate cholesteatoma heredity using Swedish registers. The research has been conducted in collaboration between Karolinska Institutet (Department of Clinical Science, Intervention and Technology and Institute of Environmental Medicine), Karolinska University Hospital and Umeå University. The research is published in the journal *JAMA Otolaryngology–Head & Neck Surgery*.

"We know little about the reasons why cholesteatoma occurs and therefore this study contributes with an important piece of the puzzle

also leading us to further hypotheses and questions. The importance of heredity is also something that our patients ask about, and although we still lack many answers this study has moved us forward," says Åsa Bonnard, [medical doctor](#) (ENT), ear surgeon and first author of the study.

More information: Åsa Bonnard et al, The Risk of Cholesteatoma in Individuals With First-degree Relatives Surgically Treated for the Disease, *JAMA Otolaryngology–Head & Neck Surgery* (2023). [DOI: 10.1001/jamaoto.2023.0048](#)

Provided by Karolinska Institutet

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