PRESS RELEASE

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Gene responsible for common hearing loss identified for first time

A gene responsible for the single most common cause of hearing loss among white adults, otosclerosis, has been identified for the first time, a scientist told the annual conference of the European Society of Human Genetics in Nice, France, today (Sunday 17 June). Ms Melissa Thys, from the Department of Medical Genetics, University of Antwerp, Belgium, said that this finding may be a step towards new treatments for otosclerosis, which affects approximately 1 in 250 people.

Otosclerosis is a multifactorial disease, caused by an interaction of genetic and environmental factors. The outcome is a progressive hearing loss as the growing bone in the middle ear interrupts the sound waves passing to the inner ear. While the causative factors remain unknown, now one of the genetic components has been identified, Ms Thys told the conference. “The gene in which the variant is located points to a pathway that contributes to the disease. This may be a lead for better forms of treatment in the future; currently the best option is an operation. However, there is often an additional component of hearing loss which can’t be restored by surgery. As the gene involved is a growth factor, and the disease manifests itself by the abnormal growth of bone in the middle ear, it may have a large potential for therapy”, she said. Improved understanding may also lead to prevention strategies.

Ms Thys and her team decided to study a gene called *TGBF1* which they already knew had non-genetic indications of involvement in otosclerosis: it plays a role during embryonic development of the ear and is expressed in otosclerotic bone. They used SNP (single nucleotide polymorphism) analysis, or looking at DNA sequence variations occurring in a single nucleotide, A, T, C or G, to study a large patient and control population from Belgium and The Netherlands. They found significant results for an amino acid changing SNP in*TGBF1*, and that this remained significant after correcting for multiple testing. Analysis of a large French group showed the same association.

“Combining the data from both groups with a common odds ratio gave a very significant result, from which we were able to conclude that we were the first to identify a gene that influences the susceptibility for otosclerosis”, said Ms Thys. “And, as further evidence, we were also able to show that a more active variant of this gene is protective against the disease.”

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