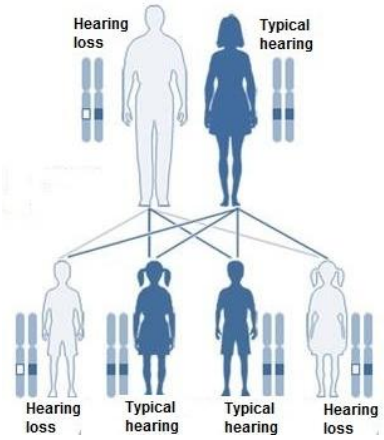
MEMORIAL UNIVERSITY
Faculty of Medicine

# Genetic Factors with Hearing Loss

**Genes** are the instructions for how our body grows and functions. Genes can be read like words in a book. If there is a spelling change in the gene, or word, it does not work properly.

***FOXL1* c.976_990del** is the name of a specific change in a gene, which changes the instructions for the function of hearing. People who inherit this genetic change are likely to develop some degree of **otosclerosis**, which is a change in the size and hardness of bone in the ear.

We typically inherit two copies of each gene, one copy from our mother, and one copy from our father.
When the inheritance pattern is **autosomal dominant,** as is seen with this genetic change, the trait is expressed if one inherited copy of the gene is not working properly. A child in this family has a 50% chance of inheriting a non-working copy of the gene and a 50% chance of inheriting the working copy.




Effects on hearing caused by *FOXL1* c.976_990del:

- Changes in hearing can start in teens up to mid-adulthood.
- Progression and severity of loss is variable, even between members of the same family.
- **Treatment**: If the bony growth is present mainly in the middle ear, surgery can be very effective in restoring most hearing. If surgery is not possible, hearing aids can be beneficial.
- Severe hearing loss may be treated through cochlear implantation.

Additional information can be found in the paper written about this specific change:

Abdelfatah, N., Mostafa, A.A., French, C.R., Doucette, L.P., Penney, C., Lucas, M.B., Griffin, A., Booth, V., Rowley, C., Besaw, J.E., Tranebjærg, L., Rendtorff, N.D., Hodgkinson, K.A., Little, L.A., Agrawal, S., Parnes, L., Batten, T., Moore, S., Hu, P., Pater, J.A., Houston, J., Galutira, D., Benteau, T., MacDonald, C., French, D., O'Rielly, D.D., Stanton, S.G., Young, T.L., 2021. A pathogenic deletion in Forkhead Box L1 (FOXL1) identifies the first otosclerosis (OTSC) gene. Hum. Genet. 1. https://doi.org/10.1007/s00439-021-02381-1

For more information about hearing loss, hearing testing and hearing loss management, contact our team audiologist Anne Griffin, at anne.griffin@med.mun.ca or (709) 489-7226 (work); (709) 293-4854 (cell)

If you have any questions or concerns about the information presented here, do not hesitate to contact our genetic counsellor, Kathy Hodgkinson, by phone: (709) 864-6694, or by e-mail: khodgkin@mun.ca

Version 1.2 December 2021