

# NEWFOUNDLAND AND LABRADOR HEREDITARY DEAFNESS NEWSLETTER

SPRING/SUMMER ISSUE

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## Introduction Dr. Terry-Lynn Young



### Study Background

Hearing loss is the most common type of disability. It accounts for 2.8 million hard of hearing and 310,000 Canadians living with deafness. Hearing impairment can be inherited or it can develop as the result of childhood infection, exposure to excessive noise, physical injury or a side effect of antibiotic use. Most hearing loss in developed countries like Canada is genetic, that is, a hearing loss caused by a change in a gene that can be passed down from generation to generation.

It became clear several decades ago that some schools in the province had large numbers of deaf students. Many of these children came from the same family or lived in nearby communities. The purpose of our research is to identify all of the genes that cause hearing loss in our province. We can then bring this knowledge to family doctors and nurse practitioners to help them better diagnose inherited deafness and refer patients for genetic counseling. Knowing the genes will provide accurate diagnosis so that treatment and education are in line with the individual's type of hearing loss.

It is clear that hereditary hearing loss is under-diagnosed in this province. Most genetic deafness is not recognized, so our list of families referred to the provincial Medical Genetics program is not complete. In 2001 we started a special effort across the province to identify families with hereditary deafness and invite them into our study. To date, we have 163 people in the study. We are currently designing new posters for doctors' and audiologists' offices to invite others to participate.

### Hello from the Recruitment Team

Welcome to the first issue of our newsletter. We plan to publish once or twice a year and keep you updated on what's happening with this research project. First of all, we'd like to thank everyone for their participation and help thus far in the study. There would be no research done without you.

There have been several team members working on this project. Some of you may have been contacted by them. Up-to-date contact numbers can be found on the second page of this newsletter. We welcome your comments and feedback.

### Meet Our Team:

Terry-Lynn Young —Principal Investigator  
Jane Gamberg —Study Leader  
Elizabeth Dicks—Clinical Scientist  
Sue Moore—Pediatrician  
Robyn Wade—Research Genetics Counselor  
Kathy Hodgkinson—Genetics Counselor  
Andree McMillan—Genetics Counselor  
Dante Galuteria—Research Assistant (lab)  
Carol Negrijn—Research Assistant—(recruitment)

On the left here, you'll see a box with a list of team members who are working on this project. We've done a short biography on three members and our students, you'll see that on Page 2. In upcoming newsletters we will showcase more team members so you can get to know them.

We've included the web site information for the Harvard Medical School Centre for Hereditary Deafness—<http://hearing.harvard.edu/>—if you have access to the internet, this site can be a valuable source of information.

Stay tuned—in our next issue we will have lots more news.

Funding for our project by:

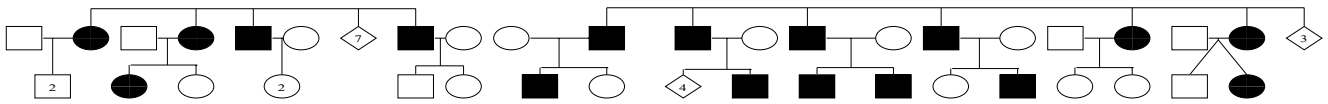


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## The Genetic Cookbook

**A Look at Genetics** by Kathy Hodgkinson

### What are Genes?

Genes are units of genetic information that pass characteristics from one generation to the next: a blueprint for how an individual will look and develop.

Genes are present in each cell of our bodies, and are passed from parent to child in a woman's egg and a man's sperm.

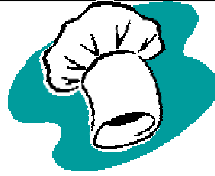
Genes always come in pairs...which is easy to remember as so do Levi's and Wranglers!! One of each pair of genes is passed from a parent to a child. The child thus gets each pair of genes back again, one of which is inherited from mum, and one from dad.

Genes are made of Deoxyribonucleic acid (DNA). DNA is a biological data set, written using a four letter alphabet: A,T,C and G.

A gene is a message, with a start, a middle and an end, and the message is written in a long string of A's T's G's and C's in a particular order. This order allows biological words to be written so the body can read the text and do what the message says.

It may be easier to think of a gene as a recipe. Like a recipe written in our 26 letter alphabet, which provides information in the form of words about how to make a cake, a gene recipe produces a message in the form of biological words that tells the body how to make a protein.

Next time: *What do Proteins Do?*



## Frequently Used Terms

**Conductive Hearing Loss**—sometimes called “mechanical hearing loss”. This type of hearing loss occurs if something is not working in the ear canal, eardrum, middle ear bones or the middle ear space. Temporary hearing losses are almost always conductive. They are far more common than permanent hearing losses. Some causes are fluid in the middle ear from ear infections, a hole in the eardrum from a bad infection or injury; narrowing of the ear canal due to surgery or disease; excessive ear wax that plugs the ear canal.

**Sensorineural Hearing Loss**—sometimes called “nerve hearing loss”. Permanent hearing losses are usually sensorineural but may also be conductive. Of the many reasons for permanent hearing loss, genetics is the most common. Other causes are structural problems; drug related; physical injuries; and infections.

If someone has both sensorineural and conductive hearing losses, it is called a **Mixed Hearing Loss**.

We have the same set of genes in every cell of our body, these genes tell us how to function and if there is a mutation (a change in the gene), it can stop the gene from working properly.

Information obtained from the Harvard Medical School Center for Hereditary

**Dr. Terry Lynn Young: Leader of gene discovery projects in Newfoundland and Labrador,** Dr. Young is a CIHR-RPP New Investigator. She recently completed postdoctoral studies at the Department of Genomics, University of Washington, USA (2000-2003). Dr. Young is currently an Assistant Professor at Memorial University (2003) and has established an externally funded gene discovery research program in Newfoundland. Her research work lead to several novel gene discoveries in Bardet-Biedl

**Dr. Jane Gamberg:** Jane has a strong research background. She has worked on a multitude of research projects over the past 25 years and recently completed her PhD, which involved studying the immune response to the human immunodeficiency virus (HIV). Jane heads up the deafness study and is responsible for most of the laboratory work for the project.

**Carol Negrijn:** Carol has recently joined the hereditary deafness team, coming from a genetics colorectal cancer study. She has been in research since 1999, after working for 15 years as a nurse on various medical units. Carol is doing recruitment and is the contact person for the study.

**Students: Nancy Merner** (a PhD student), **Sandra Cooke**, and **Lance Doucette** (both Masters students) have all participated in the deafness study as part of their graduate school projects. We expect additional students to become involved with this research over time.

**Depending on the cause, hearing loss may be temporary or permanent**

## Contact Information



Please feel free to contact us with your questions, comments and suggestions.

If you have moved or are moving or wish to receive mail at an alternate address, please write or call us. We would appreciate comments or suggestions for future newsletters.

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