

# NEWFOUNDLAND & LABRADOR STUDY ON DEAFNESS AND HEARING LOSS

FALL/WINTER ISSUE

December 2007  
Volume 2

## Carol's Corner

Welcome to the second issue of our deafness and hearing loss newsletter. We'd like to take this time to thank you for all your help in our research and to wish you all the best of the season. Our study is busily humming along, we have lots of new people involved and exciting things are happening.

Congratulations to Robyn Wade, our genetics counselor, who was accepted to medical school this past September. Our loss is her future patients' gain as she will make an excellent physician. The genetic counselor who some of you may be familiar with, Andree McMillan, has moved into another area and is no longer working with people in our hearing loss study. We are recruiting a genetic counselor for this project.

For any of you out there who have forms to be filled out, it's not too late, and we're as close as the telephone or e-mail if you have questions or need help. If you happen to have a Harvard questionnaire that you are struggling with, please just fill out the family information on the last page and send it back to us - we are in the process of making that questionnaire more user friendly and will mail you the newer, shorter version when it's been approved by our ethics committee.

Happy Holidays to you and yours in this festive season. Keep up the good work, we can't do research without you.



Courtesy of Thomas Clench

*Seasons Greetings to you and your loved ones*

## What's New in the Lab? by Jane

My name is Jane Gamberg and I'm responsible for the laboratory portion of the study. The experiments required to identify mutations in genes involve many steps. Because some of you might be interested, I thought I'd explain a little about the work we do in the laboratory.

I'll start at the beginning; we receive the blood samples that many of you so generously donate, and the first thing we do is separate the DNA from the rest of the blood components. This is because almost all of our testing is done using DNA. The procedure is simple and results in the DNA forming a 'clump' that can be separated from the rest of the liquid. Once we have the DNA, we store it in tubes in large refrigerators. Scientists like working with DNA because it can remain stored for long periods of time without becoming damaged. Then, as new information is discovered, a portion of the DNA can be removed and tested.

We have already started testing your DNA samples for several of the genes that are known to cause hearing loss and, for some families, the cause may be discovered quickly. However, it will probably take a long time to find all the answers because there are many genes involved in the hearing process and we're only just beginning to find them and understand how they work. I'll tell you more about that in the next issue. For now, the laboratory personnel would like to thank you all for your continued support (keep those blood samples coming!), and we wish you and your families a safe and happy holiday season. All the best for 2008!



## Frequently Used Terms

**Autosomal dominant** - A mutation in a gene that is strong enough to make a person affected even if the person also has a normal copy of the gene.

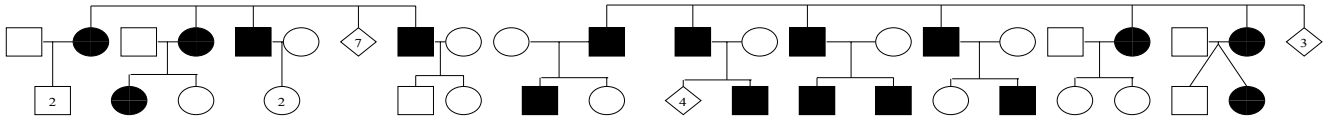
**Recessive** - A mutation in a gene that is not strong enough to make a person affected if the person also has a normal copy of the gene.

**X-Linked** - A special form of inheritance that involves mutations on the X-chromosome.

*Information obtained from the Harvard Medical School Center for Hereditary Deafness web site.*

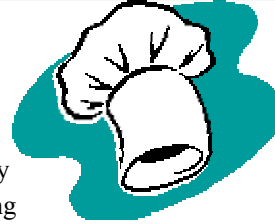
Funding for our project by:





## The Genetic Cookbook

A Look at Genetics by Kathy Hodgkinson



### What do proteins do?

Proteins make up all the cells in living tissue. Cells are very important as they perform the jobs that keep each tissue and organ system in our bodies working properly. If a gene recipe is changed, the protein it makes will also be changed. The result may be too much, or too little protein. This may mean that the protein cannot do its job properly.

### Why does a change in a gene (and therefore a corresponding change in a protein) matter?

If the change in a gene is small, it may not matter. The protein can probably still do its job. If however the change is more significant, the protein cannot do its job.

If we go back to our cake recipe analogy, we can think of the gene recipe as the cake recipe, with the resulting protein as the cake. If a cake has four extra tablespoons of flour, it will still look and taste like a cake, so it can still be used for the birthday party.

However, if the flour is missing from the recipe entirely, then the cake will NOT look, nor will it taste like a cake, and the birthday party will be spoiled.

### What are disease genes?

These are genes that because of the recipe change within them, make a protein, that like the cake without the flour, cannot work properly. When a protein cannot work, it cannot do its job, and the cell will not function. This can have a devastating effect on the tissue and the organ systems to which the cell belongs. Using the same analogy as before, the cake is inedible, and the party is spoiled.

### How do you know when a disease is caused by a problem within a gene?

When a disease runs through families in a particular pattern, people who are used to looking at genetic disease can usually tell if the disease is caused by a mistake in the recipe of a single gene. Sometimes we cannot tell.

.....more next time.....

### Did you know?

- While there are families that have hearing loss in many generations, some people with genetic hearing loss do not have family members with hearing loss.

- Poking objects too deeply into the ear canal (Q-tips, pencils, food, buttons, pins, etc) can cause hearing loss; it is the most common ear injury in toddlers.

*From Harvard Medical School Center for Hereditary Deafness*



## More Biographies:

**Elizabeth Dicks** - Elizabeth (aka Betty) is a Clinical Scientist for Genetics at Eastern Health and is responsible for all of the clinical aspects of the genetic studies presently being conducted. She is also the voice on the phone when you call our 1-888 toll free number.

**Dante Galuteria** - Dante is a research assistant in Terry's lab. He has an extensive genetics background, he isolates DNA from our hearing loss samples as well as from samples for all other projects in the lab.

**Kathy Hodgkinson** - Kathy is a genetics counselor with Eastern Health, is a PhD candidate and works on various genetics projects. She is the author of our Genetics Cookbook section.

**Sue Moore** - Sue is well known at MUN for her work on Bardet-Biedl syndrome and has now completed a residency program in pediatrics in the Faculty of Medicine. She is participating in the hereditary deafness study as she sees many hearing impaired children in her practice.

**Anne Griffin** - Anne is a clinical audiologist, and has worked at Central NL Regional Health Centre for the past 18 years. She does hearing testing on all age groups to identify hearing problems and solutions, and provides rehabilitation services for hearing problems which cannot be treated medically or surgically. As an expert member of our research team, she reviews the hearing history of family members and interprets the types and patterns of hearing loss in each individual and family.

## 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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