

Kolling Correspondence

1 IN 500 AUSTRALIANS FOUND TO HAVE A GENETIC CAUSE OF HEARING LOSS

Researchers at Sydney's Kolling Institute of Medical Research have discovered that a genetic mutation, known to cause hearing loss affects 1 in 500 Australians.

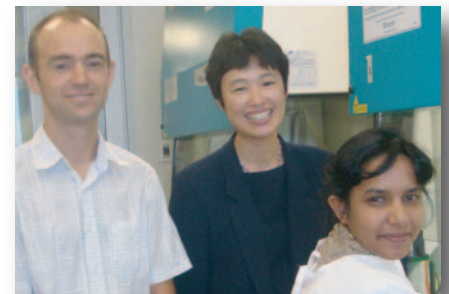
The genetic mutation (known as the m.1555A>G mutation) was previously thought to be quite rare; however, a new study published in the prestigious New England Journal of Medicine this month showed that more Australians are affected than originally thought. Individuals who have this genetic mutation are at risk of developing hearing loss, particularly if exposed to some types of drugs commonly used to treat serious infection e.g. antibiotics including neomycin, gentamycin and streptomycin. Exposure to even small doses of these drugs may cause hearing loss in affected individuals

Scientists in the Department of Neurogenetics at the Kolling Institute carried

out genetic analysis on nearly 3000 subjects and found that six of these people had the m.1555A>G mutation. All the carriers of the mutation had developed hearing loss although none realised that their hearing loss was due to a genetic mutation. Only three of the subjects had a family history of hearing loss. The m.1555A>G mutation, is found in a part of the cell called the **mitochondrion - the cell's powerhouse** (illustrated above left). The mitochondrion acts as the "battery" for the cell, providing most of the cell's energy. Mitochondria are found in almost every cell in the body, but if the mitochondria do not supply energy to the cells of a particular organ or tissue, symptoms affecting that organ may develop.

Patients with mitochondrial genetic mutations may develop symptoms of hearing loss, muscle weakness and fatigue, diabetes, stroke-like episodes or seizures. Although patients are born with mitochondrial genetic mutations, they may not develop symptoms until later in life. Furthermore, because

mitochondrial disease has been perceived as a rare disease, diagnosis of this condition is sometimes delayed.



Kolling researchers Dr Neil Manwaring, Dr Carolyn Sue and Dr Himesha Vandebona

The inaugural "Mitochondrial Information Day" was held at the Kolling Institute in March 2009.

For further information, please contact Dr Carolyn Sue or Wendy Welsh on 02 9926 6491.

KOLLING FOUNDATION REVITALISED

Kolling Foundation is delighted to announce that its new management team of CEO Sean Wilson and Director of Communications Suzie Freebury is now up and running.

Kolling Foundation serves a dual purpose:

- **Raising funds to support and assist medical and scientific research** at the Kolling Institute of Medical Research plus other groups on the RNSH Campus;
- **Promoting and increasing awareness and understanding of the importance of medical research** at Kolling, across the community at large.

Over the next few months we will be delivering a fresh new look and feel for Kolling – a new brand, a new logo and an updated newsletter.

We'll let you know how you can support the great work here at Kolling and we'll be looking for your feedback too.

Visit the Kolling Foundation website today!
www.kolling.com.au or phone 02 9926 4904
To donate: www.kolling.com.au/donate

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