

September 2002
EDITION 5

Our Mission

To promote and develop awareness of genetic conditions within the community, provide information and advocacy to individuals and families, foster research, encourage and support service delivery by genetic support groups.

Welcome to the September 2002 edition of the Genetic Support Council Western Australia (Inc) newsletter.

Current Issues and matters of interest

Autumn 2002 – Volume 18, Number 3 Epilepsy & Genetic Research

Canada

In May, researchers at the McGill University Health Centre (MUHC) in Montreal announced that they have identified a gene underlying juvenile myoclonic epilepsy.

Previously, a few mutated genes have been shown to be associated with rare forms of epilepsy. However, the variability in clinical features of more common epilepsies has hampered the search for genetic bases of these syndromes.

This collaborative study, led by Dr. Guy Rouleau (MUHC neurologist, and professor of neurology and neurosurgery at McGill University), co-authored by Patrick Cossette, and including researchers from the Université de Montréal and University of Toronto, analysed DNA from 14 members of a French-Canadian family, 8 of whom have JME. In those 8, researchers found identical mutations in a well-characterized gene, GABRA. This

gene encodes the alpha 1 subunit of the GABAA receptor (a chloride channel) which is involved in neuronal transmission. Research confirmed that this mutation alters the function of the GABAA receptor: the mutant protein has diminished activity in cultured cells. This results in abnormal electrical patterns in the brain. There are 16 different subunits which can contribute to a functional GABAA receptor in the brain. Previous research shows that mutations in a different subunit of the GABAA receptor are involved in fever-induced seizures. This study, in which the GABAA receptor is implicated in a distinct and more common seizure disorder, suggests that this molecule may be centrally involved in epileptic syndromes. Screening families for mutations in each subunit may yield further insight.

Most people living with epilepsy receive anti-seizure medication therapy throughout their lives, which may cause severe side effects. Identifying and characterizing the genes which cause epilepsies may help in designing more specifically targeted drugs for the treatment of these disorders.

Australia

The research team led by Josef Gecez, Department of Cytogenetics and Molecular Genetics at the Women and Children's Hospital (Adelaide, Australia) reports that it has discovered a gene linked to epilepsy and learning disabilities.

They report that the gene affects the way the brain works by controlling how, where and when other genes function. A mutation in the gene can produce a number of different conditions, including epilepsy, infantile spasms (West syndrome), dystonia (a disorder of muscle tone) and learning problems. The gene, on the x-chromosome, is one of about 100 genes on that chromosome that can affect IQ.

Genetic material for their studies came from 9 families in Australia, Belgium, Canada, and Norway.

Sources

<http://epilepsyontario.org>

<http://www.newswire.ca/releases/May2002/09/c3610.html>

<http://genetics.nature.com>

<http://www.efa.org/epusa/061702/hunt.html>

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The views expressed in this newsletter are those of the individual authors and not necessarily those of the GSCWA. The material supplied is for information purposes only, and is not to be used for diagnosis/treatment.

...more matters of interest

At the request of the federal government, the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) are conducting a joint inquiry into the protection of human genetic information. The ALRC and AHEC have been asked to consider what type of regulation may be needed, in relation to human genetic samples and information to:

- Protect privacy;
- Provide protection from unfair discrimination; and
- Ensure high ethical standards of conduct.

Discussion Paper 66 was released on 28 August 2002. For more information log on to www.alrc.gov.au.

The Executive Summary as well as their proposals and questions can be reviewed at our website under "**Publications**".

The outcome of the forum on the discussion paper being held on the 23 September at Grace Vaughan House will also be published on our website as soon as it becomes available.

If you have any enquiries or want to comment on the discussion paper, do not hesitate to contact us. We value your opinion!

Coming Events

Genomics, Society and Human Health, Institute of Advanced Studies, UWA

You are invited to a public lecture by
Professor Ruth Chadwick

Professor of Bioethics and Director

Institute for Environment, Philosophy and Public Policy

Lancaster University

"Genetic Possibilities, Information And Choice"

Geography Lecture Theatre 2, The University of Western Australia

Tuesday 1 October at 6.00pm

The argument that consumers, whether of health care services or anything else, should have choice, is one of the arguments most commonly advanced, and upheld as if it were unproblematic. Choice is upheld as an ideal about the medical applications of genetics – whether to take a test; what action to take following genetic counselling. It is important, however, to have regard to the context in which choices are made. A number of factors should be taken into account, including not

only the boundaries to the range of options available, but also more general features contemporary social context.

One of these features is the emphasis put on what information is necessary in order to make an *informed* choice. In the context of genetics, however, it has been argued that there may be something special about genetic information which makes the notion of informed choice particularly complex. While it may not be true that genetic information is different in kind from other sorts of medical information, decisions that now confront individuals, such as whether or not to contribute DNA samples to a database, or whether to undergo pharmacogenetic testing, raise issues of uncertainty and unpredictability of long term consequences. This complicates the ethical issues in such a way that it is unlikely that they can be solved by reliance on choice alone.

Source:

<http://www.gshh.uwa.edu.au/events.html>

**Genomics, Society and Human Health,
Institute of Advanced Studies
and**

**Institute for Sustainability and Technology
Policy, Murdoch University**

**Present a Cross Disciplinary Symposium in
October 2002**

"Being human in an age of genetics"

Convened by

Dr Beverley McNamara, Anthropology, UWA
Dr Ian Barns, Institute for Sustainability and
Technology Policy, Murdoch University

October 2 all-day symposium

The extraordinary advances in genetic research and technologies have stimulated lively debates about the role of science in our lives. These debates usually focus on the legal and ethical implications of emerging genetic technologies. Much less attention is given to what the new genetics means for our understandings of personhood. This symposium addresses this very basic question: what does it mean to be human in an age of genetics? Presentations by three experts in key areas of genetics and society will set the scene for a panel discussion of some very different perspectives on the meaning of genetics for human personhood

The symposium will include contributions from scientists, social scientists and humanities scholars including:

Dr Christopher Newell, AM, School of
Medicine, University of Tasmania
Professor Ruth Chadwick, University of Central
Lancashire

Dr Ian Barns, Murdoch University

Dr Beverley McNamara, UWA

Professor Alan Bittles, Centre for Human
Genetics, Edith Cowan University

And will consider the following questions:

- What challenges do humans face in an age of genetic technology?
- Genetics and humanness.
- Will developments in genetics change what it means to be human?
- The idea of 'redesigning' humans
- The state of play: what is happening in genetic research today.
- A global view: what challenges do humans face in an age of genetic technology?

To register interest in attendance, please contact the Institute of Advanced Studies, UWA, 9380 2114/1340
tawhite@cyllene.uwa.edu.au

Source:

<http://www.gshh.uwa.edu.au/events.html>

The Law School and the Genomics, Society
and Human Health program at
The University of Western Australia
Have much pleasure in presenting the

John Toohey Oration

***"Genomics and Democracy - A Global
Challenge"***

Delivered by

Justice Michael Kirby

At 8pm on Thursday 24 October 2002
Social Science Lecture Theatre, UWA

"Justice Michael Kirby will be honouring his distinguished past-colleague on the High Court, Justice John Toohey in the John Toohey Oration."

In this Oration, Justice Kirby will explore a number of issues that have arisen in some of the international activities in which he is involved. He was recently reappointed to the International Bioethics Committee of UNESCO. He also serves as a Member of the Ethics Committee of the Human Genome Organisation and as Honorary Adviser to the High Commissioner for Human Rights on Bioethics.

Some of the most acute bioethical problems are presented by the use that is now being made of knowledge about the human genome - the map of the genes of our species. Several puzzling legal issues are arising that need to be solved by national legal systems. These include:

- The protection of privacy of genomic data;
- The use of that data by employers and insurers;
- The patenting of genomic sequences; and

- Benefit sharing to ensure that the genome is "about malaria, not just wrinkles".

Justice Kirby promises that the science will not be too complicated. He is not a scientist. But he cannot make the same promise about the ethical choices that have to be made. Ensuring that such problems are addressed in an informed and participatory way is a major global challenge for democracy and international, as well as domestic, law.

Source:

<http://www.gshh.uwa.edu.au/events.html>

More events:

September 2002

01.09. – 07.09.2002

Spina Bifida Awareness Week

for more information, call 9389 8311

08.09. – 14.09.2002

Parkinsons Awareness Week

Parkinsons Association

for more information, call 1800 644 189

23.09. – 29.09.2002

WA Stroke Week

WA Strook Foundation

for more information, call 9272 1122

October 2002

01.10. – 31.10.2002

Lupus Aqareness Month

The Lupus Group of WA

for more information, call 9224 3144

06.10. – 12.10.2002

Mental Health Week

WA Assn. for Mental Health

for more information, call 9420 7277

13.10. – 19.10.2002

Foot Health Week

Australian Podiatry Assn.

for more information, call 6398 0225

20.10. – 26.10.2002

National Carers Week

Carers WA

for more information, call 9424 5210

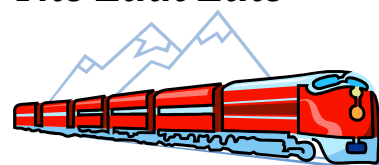
A letter

Mandy - Trisomy 18

This letter raises concerns about services to children within the education system. The letter focuses on the need for changing educational approaches for children with special needs and calls for parents to come forth to participate.

To view the whole letter, see our website <http://geneticsupportcouncil.org.au> under "Current Issues"

The Link Line



The Link Line provides a supportive and confidential means of connecting individuals and families for whom no known genetic support group exists.

If any individual is seeking contact with others in these circumstances, *The Link Line* is available to you for this purpose.

Genetic support groups are an important resource for families or people in a similar situation. To date, there appears to be no specific support group for the condition Ehler Danlos (connective tissue disorders) in Western Australia. A family living in the southwest region of Western Australia is seeking linkage with other people and families living with this condition.

To make confidential contact with anyone using *The Link Line*, or for further information regarding this service, please call Terry Keating at GSCWA.

Advertise in 'Gene Advocate' or on our website!!!

Have you got anything you want to be published? Either our newsletter or our website can be the place to display your information. So contact us via e-mail or phone and we can arrange it for you.

Interesting Websites

Assistive Media

<http://www.assistivemedia.org/>

A free audio-literary service for persons with print reading/access barriers.

- The mission of Assistive Media is to heighten the educational, cultural, and quality-of-living standard for people with disabilities and help achieve independence and become better integrated within the mainstream of society and community life in general. Assistive Media accomplishes this by providing free-of-charge, copyright-approved, high caliber audio literary works to the world-wide disability community via the Internet effectively, inexpensively and efficiently.

Pediatric Neurological Associates; USA.

<http://www.pediatricneurology.com/>

This Web site is sponsored as a resource to patients by Pediatric Neurological Associates; USA. It provides information about pediatric neurology on childhood seizures, epilepsy, headaches, migraine, autism, Asperger's syndrome, PDD, pervasive developmental disorder, developmental delay, tics, child brain tumors, Tourette's syndrome, types of learning disability, ADHD, ADD, simulations of Attention Deficit Hyperactivity Disorder, medications such as Ritalin, EEG tests, and MRI tests and an online ADHD book. The 'Best Bets' page can lead you to any other topic in child neurology.

Health On the Net Foundation

<http://www.hon.ch/home.html>

The Health On the Net Foundation (HON), created in 1995, is a not-for-profit International Swiss Organization. HON's mission is to guide lay persons or non-medical users and medical practitioners to useful and reliable online medical and health information. HON provides leadership in setting ethical standards for Web site developers. It contains:

- The MedHunt database includes a complete list of hospitals on the WWW, Internet medical support communities (listservers, newsgroups and FAQs) and extensive internet WWW resources for healthcare providers, medical professionals and individuals.
- MedHunt proposes a spell checker, a list of medical/health topics for a given query, the MeSH terminology (a hierarchical structure of 18,000 medical concepts from the National Library of Medicine), and a display of the results by geographical, domain name, and ranking preferences.
- MedHunt is devoted to discovering medical information on the internet by way of M.A.R.V.I.N. Marvin, a robot that searches sites and documents specifically related to a given specialised field.
- HONselect, HON's new search integrator for strictly medical and health queries. HONselect combines five information types - MeSH® terms, authoritative scientific articles, healthcare news, Web sites and multimedia - into one service to focus and accelerate your search. It is also available to search in other languages.

Special Children Disability Links

<http://www.irsc.org:8080/irsc/irscmain.nsf>

This site was developed by Julio Ciamarra when his son was born with multiple disabilities, and he found that information on children with disabilities was difficult to find. Ciamarra's profession is working with Internet technology.

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Alzheimer's Association of WA
Angelman Syndrome Association
Australian Huntington's Disease Association
Australian Pituitary Foundation WA Branch
Australian Tuberous Sclerosis Society Inc.
Coeliac Society of WA
Cushings Disease Support Group
Cystic Fibrosis WA
Dyslexia SPELD Foundation WA Inc
Epilepsy Association of WA
Haemophilia Foundation WA Inc.
Heart Kids WA
Klinefelters Support Group
Learning and Attention Disorders Society of WA (LADS)
Mucopolysaccharide & Related Diseases Society (MPS)
Neurofibromatosis Association of WA.
Parents of Children with Disabilities
Short Statured People's Association WA Branch
SIDS and Kids Western Australia
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Thalassaemia Association of WA
The Periodic Paralysis Society of Australia
Trisomy and Related Disorders
Western Australian Retinitis Pigmentosa Foundation
WA Tourette Syndrome Organisation (WATSO)

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