



# Snippets from the Centre

MARCH 2010

Welcome to the inaugural edition of the Centre for Genetics Education's newsletter, *Snippets*.

We've long believed in servicing NSW with informative and trustworthy genetics communication tools and now we're happy to add a quarterly newsletter to our service. We aim to fill it with news regarding our latest activities, research and resources that we have created. It will also be an opportunity to share recent genetics media and some light-hearted entertainment such as movies and books on our favourite topic, genetics!

We hope this will be an opportunity for you to get to know us and come to understand how we can help you or your genetics service.

So, please, take a break from your day for a minute or two and discover what has been keeping us busy this year so far!

**Jacinta Reid**  
*Health Education and Communications Officer*

## Resources ready for you

**We are always adding to the great suite of resources we have available for you.**

Our information sheets, regarding the rarer and complex genetic conditions, are updated upon request. Some of the most recent ones updated include:

- Kabuki syndrome
- Oculopharyngeal muscular dystrophy
- Branchio-oto-renal (BOR) syndrome
- Spondyloepiphyseal dysplasia congenita (SEDC)
- Pyruvate dehydrogenase deficiency (PDD)
- Batten disease
- Cardiofaciocutaneous (CFC) syndrome
- Carbamyl phosphate synthetase (CPS)

To request an information sheet, please email us at [contact@genetics.com.au](mailto:contact@genetics.com.au)

## New look for the Centre

**This year the Centre will be celebrating its 21<sup>st</sup> birthday and to mark the occasion we've adopted a new look and a new logo.**

We can now be recognised by bold blue colours and stripes (as used in this newsletter at the top and bottom of the page) and an exclusively designed 'DNA man' takes pride of place as our logo.

Our website has also been redesigned to reflect our new look and contains some great new features:

- A genetic conditions support group list
- A pregnancy preparation section.

Don't worry though; we've kept some favourites such as the Fact Sheets and multilingual resources.

[www.genetics.edu.au](http://www.genetics.edu.au)



## Latest published research from staff at the Centre

*Journal of Genetic Counseling* Volume 18, Issue 5 (2009); 436.

### **Challenges Faced by Genetics Service Providers' Practicing in a Culturally and Linguistically Diverse Population: An Australian Experience**

Mona Saleh, Kristine Barlow-Stewart, Bettina Meiser, Ian Muchamore

This paper showcases how clinical genetics services are facing and overcoming some of the challenges presented when counselling patients from different cultures.

Through the cooperation of 53 clinical genetics practitioners across NSW and Victoria, seven focus group sessions were held to document some of these challenges and how they were being addressed. The discussions explored participant's experiences and definitions of cultural diversity; their use of educational resources with clients; their experiences working with interpreters; and the impact culturally specific education/training had had on their practice or confidence with culturally diverse clients.

As diverse as the specialty and physical locations of the professionals were, so too were their responses towards the topics of discussion. Ranging from one participant who told of counselling ten non-English speaking clients in a week to another who believed they hadn't used an interpreter in 12 years of practice.

Resulting from the focus groups and by breaking down the discussion points into nine themes and 23 subcategories, areas of challenge and recommendations were established and detailed. Among the findings the most significant was the need to think about culture as a concept beyond geographical or ethnic origins. In addition, the need for more current and accurate multilingual information for clients was identified.

To view the resources developed as a result of this research visit [www.genetics.com.au/multilingual.asp](http://www.genetics.com.au/multilingual.asp).

To obtain a full copy of the paper please email [msaleh@nscchahs.health.nsw.gov.au](mailto:msaleh@nscchahs.health.nsw.gov.au).

***Mona Saleh is a Manager and Genetic Counsellor with the Centre for Genetics Education. Mona is currently completing her PhD.***

### ***Reproductive Medicine Online***

#### **Psychological impact of preimplantation genetic diagnosis: a review of the literature**

JC Karatas, KA Strong, K Barlow-Stewart, C McMahon, B Meiser, C Roberts

This review begins with a summary of the known psychological aspects of IVF, followed by a review of the limited available literature exploring the psychological and broader psychosocial impact of PGD.

Through a selection and exclusion process, 19 studies were deemed as relevant for the literature review. The studies included attitudinal surveys of non-PGD users, and retrospective and cross-sectional studies with PGD users. Psychological adjustment and well-being were a small component of two studies exploring the development of children born after PGD at two years postpartum.

Overall findings suggested that those with a significant history of traumatic reproductive experiences were more likely to perceive PGD as a possible reproductive option. Additionally, PGD was reported to be 'extremely stressful' and 'emotionally draining'.

The findings confirmed that there was a need for additional research in this developing area, with a focus on prospective studies using validated psychometric scales, that explores anxiety and depression at various time-points through the PGD process, as well as the impact of PGD beyond the establishment of a pregnancy, parent-infant attachment, and long-term follow-up of women for whom PGD was unsuccessful.

To obtain the full paper please email [jkaratas@med.usyd.edu.au](mailto:jkaratas@med.usyd.edu.au).

***Janan Karatas is a PhD student with the Centre for Genetics Education, submitting her thesis in March.***

## Media Spotlight on the Centre for Genetics Education



The Centre for Genetics Education is continuing to prove the 'go-to' source for hot genetics stories, as was the case in late February when news broke of a new genetics service being offered by NIB Health Insurance.

The *Sydney Morning Herald* reported that 5000 NIB members were being offered genetic testing through Navigenics. The testing would search for risks relating to illnesses such as diabetes, heart attacks and some cancers. However, concerns were raised about the implications of genetic testing results on life insurance, which is also offered by NIB.



An NIB spokesperson told the *Sydney Morning Herald* that "customers were warned of the implications in fine print on the letter saying 'you may be required to disclose genetic test results, including any underlying health risks and conditions which the tests reveal, to life insurance or superannuation providers'".



The Centre's Director, Associate Professor Kristine Barlow-Stewart was called upon to outline the rights and responsibilities for people who undergo genetic testing and also those of life insurance companies.



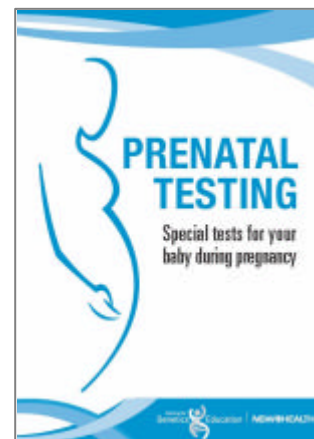
Many radio stations, newspapers and TV newsrooms called for some insight and interviews, including *The Age*, *The West Australian*, ABC TV, ABC Radio (metro and regional stations), 2GB, *The 730 Report*, *Hungry Beast*, 4BC, 2HD Newcastle, *Sun Herald* and *Medical Observer*.

## Updated resources

The Centre has been busy the past few months updating two great resources to help patients and their families.

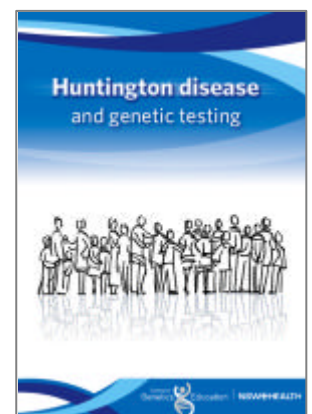
### Prenatal Testing: Special tests for your baby during pregnancy

The most requested book at The Centre has been updated with all of the latest evidence-based information regarding prenatal testing. It includes an improved pregnancy timeline, easy to understand information and pages for notes about the expectant mother's pregnancy.



### Huntington disease and genetic testing

What was once a series of brochures has now been revamped into one informative booklet. It offers excellent information on understanding the huntingtin gene, how Huntington disease can affect a family and detailed information on understanding test results.



**Both of these booklets will be arriving in our office shortly for distribution. To order a large quantity, please contact our office or keep watching the 'What's New' section on our website for a PDF version soon!**

## Genetics Media

### ➤ Parents pass on less genetic faults than first thought

Researchers from the Institute for Systems Biology in Seattle and the University of Utah have discovered that parents pass on less faulty genes than previously believed. Apparently, instead of the suspected average of 75 genetic mutations per parent, it is only around 30 per parent.

### ➤ deCODE reborn

deCODE genetics has emerged as a newly-financed, private company. deCODE's official statement said they will continue to offer the same personalised genetic services as well as a new venture into genetic-based therapies. deCODE claimed bankruptcy late last year.

### ➤ Prenatal testing at five weeks

Dutch researchers have developed the world's first prenatal test that can be performed as early as five weeks. The blood test can determine the baby's sex and detect genetic abnormalities. Research shows it is almost 100% accurate.

### ➤ Green sheep

In a bid to cut greenhouse gas emissions produced locally, Australia's Sheep Co-operative Research Center is hoping to breed burp-less sheep. Apparently the agriculture sector is the nation's second largest source of gas emissions and sheep are known to burp large amounts of methane. Scientists will study over 700 sheep from 20 different genetic lines.

## DNA Blockbuster



**With buzz words like "genetic testing", "cancer" and "survivor" always setting the scene for a good story, Hollywood will capture a non-fiction, 'genetic-based' story on film.**



The Oscar winning producer and cinematographer team from the film, *Monster*, will this year begin shooting the tales of three-time cancer survivor, Anne Parker, and American geneticist Mary-Claire King, who dedicated her professional career towards discovering the BRCA genes.

The film, to be titled *Decoding Annie Parker*, will capture the power of belief and determination with both women believing that breast and ovarian cancer was hereditary, even when peers and professionals told them they were wrong.

Ms Parker, 58, has survived breast, ovarian and liver cancer but saw her mother and sister succumb to the disease in the 1970s, both at young ages. Long before scientists knew of the BRCA genes, Ms Parker believed there was a reason why breast and ovarian cancer had such a strong presence in her family and is now an advocate of genetic testing.



"I have come to the conclusion cancer is my life. I had to make the decision for the world, for other women. I want other people to know," Ms Parker said.



## Share the spotlight

If you have news you would like to share with your peers, send your information to Jacinta Reid [jacreid@nscchahs.health.nsw.gov.au](mailto:jacreid@nscchahs.health.nsw.gov.au).

Please note there are editorial restrictions and all work will undergo editing where required. Maximum 300 words.

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