GENETIC SUPPORT NETWORK OF VICTORIA

Perspective

(Formerly GSNV News)

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Picnic in the Paddock

The GSNV and VCGS can form a strategic alliance

Over the past few months I have had the opportunity of working with A/Prof Agnes Bankier, the newly-appointed Director of the Victorian Clinical Genetics Services, on many exciting projects. These include the National Approach to Public Health Genetics paper and a working group looking at the opportunities around the development of a Centre for Community Genetics. Working with Agnes has increased our understanding of the VCGS and in turn has increased their knowledge of our role.

It is important that the GSNV is constantly working toward helping others in the community affected by a genetic condition to find the appropriate support group in order to receive information, support and advocacy. It is also important the GSNV acts as a voice to ensure the community has a say in all aspects of the ever-increasing genetic debate. In order for this to happen we need to be constantly working with others to develop strategic partnerships that support but do not dominate each other. Working together will keep us in touch with the issues, and how to best address them for you, our members.

The GSNV will be having a picnic at Hay's Paddock in Kew as part of Genetics Awareness Week on Saturday the 16th of June. This will be a perfect opportunity for all our members to meet Agnes and others from the VCGS. It's great to put faces to names. Having said that, I will hand over the letter to Agnes to introduce herself further and the role of the VCGS.

I welcome this opportunity to write to you, together with Dani, President of the GSNV. Dani and I now work on a number of projects together and share the same vision – the importance of supporting personal choice in genetics, as in other aspects of life. Well being and health are not necessarily the absence of disability or challenge. It comes from well supported, well informed life choices. It recognises the challenge and lives each day to the full.

The VCGS has a charter form the DHS to provide genetic counselling and diagnostic services to the people of Victoria. Our services are recognised for excellence internationally but unfortunately in our own state many people of Victoria and many of the doctors they turn to, do not know what is available through our services. We are in a pivotal position to be of help. We can help interpret new information from research as it reaches you from the media and the internet or when you seek our services. The availability of new tests has raised

concerns about eugenics - concerns that the new technology will be driven by a wish to eliminate birth defects. Genetic testing is about supporting informed choice and the options available or not just about testing in pregnancy.

Increasingly genetic tests are becoming available which provide information about future health of people, information about conditions they may develop at an older age. The new genetics will make it possible to develop a susceptibility profile and to do something about it. Pharmacogenetics is an emerging body of knowledge which will make it possible to offer treatment based on a persons genetic profile before the onset of disease symptoms. The VCGS is not about eugenics but about information and choice. We can also help ensure that you are linked to other services that may be of help. We cannot do this alone.

The VCGS together with the GSNV can form a strategic alliance, which can help both groups achieve what neither could achieve alone. An alliance may help gain resources from funding bodies, develop policy and provide information and support. The VCGS helps with diagnosis but ongoing care (with specific exceptions) is provided by a variety of doctors. Time and time again we hear of people's frustration with the delays in getting appropriate help. As a first step we have developed a Resource Check List which is included in this newsletter. We would value your input. Please share your knowledge and experience. Once finalised this could be made available through the GSNV, VCGS and through the respective web sites.

The VCGS is considering a name change and I thank those of you who have already responded to an initial survey of your views. Our new name will be launched just before the GSNV Picnic. You can contact me by letter or email on bankiea@cryptic.rch.unimelb.edu.au

Yours sincerely

Associate Professor Agnes Bankier
Director,
Victorian Clinical Genetic Services

Danielle Blanden
President,
Genetic Support Network of Victoria

NZORDS

New Zealand Organisation

Mark **Saturday 16 June** in your diary as a fun day for your family. The Genetic Support Network of Victoria will be hosting a picnic for all our families at Hay's Paddock. More information will be in your mailbox soon.

Geneticisation: Where Do We Draw The Line?

Dr Tom Shakespeare

Tom Shakespeare is currently research development officer at the Genetics Policy and Ethics Research Institute - International Centre for Life in England. He was formerly University Research Fellow at the Centre for Disability Studies, School of Sociology and Social Policy at the University of Leeds in the United Kingdom. This Centre has a national and international reputation built on pioneering work on the social creation of dependence and institutional discrimination against disabled people. Tom Shakespeare has published numerous journal articles and book chapters on issues surrounding disability and the definition and understanding of disability. He is co-author of The Sexual Politics of Disability (1996). Tom visited Australia earlier this year and spoke to the staff of Genetic Health and the Murdoch Children's Research Institute. Following are some highlights from this presentation.

"…I think that there are two problems which I think are salient to what we all do and how we work and think, and they are:

First of all, the tendency in James Watson's* work to reduce everything to molecular biology levels. We know this is what the guy does. He said once I think that "The only important science is physics, everything else is social work." Making a sort of reduction at this point that it all collapses into things at the levels of molecules and biochemistry. And I am impressed that your director is a chemist, so maybe I have come to the wrong place to say this, but I think he is wrong! (Not Bob♣ – James). So, "there is only physics, everything else is social work" - the reductionist mentality. He has also said that in the past people used to think that the future was written in the stars, now we know in large part it is written in our genes. So the determinist fallacy.

So two errors there, I think, in terms of the way most scientists or clinicians work (I hope, at any rate) and in terms of the that way we should understand human differences.

After all, you would not say, as you go home to your partner or friend or whatever, "That was a very good talk from somebody with a G to A transposition of point 38 of the FGFR3 gene." You wouldn't say that, I feel sure, even the clinical geneticists amongst you. You may have made a mental note "FGFR3 a bit dodgy" but you wouldn't have actually labelled me on that basis. You would have said, "Who's that bloody Pom who thinks he knows what he's talking about?"

That is to say, my molecular base change is not the salient thing about me. That's not why I am here, that's not why you are staring at me in strange ways, it doesn't explain my family, it doesn't explain my career, it doesn't explain anything about me that is of any interest. My class, my gender, my particular personal biography will explain a lot about me, my G to A transposition explains very little.

So at levels like this I think that scientists like James Watson are perpetuating what Abby Lipman♦ has called "geneticisation". The tendency to explain complicated social and indeed personal processes by virtue of (at some levels) very simple genetic or other metabolic changes. That's what I think he got wrong.

What he also got wrong, James Watson, and what I think John Harris♠ was getting wrong is

* Dr James Watson was one of the co-discoverers of the structure of DNA and is a Nobel Prize-winning molecular biologist.

♣ Professor Bob Williamson, Director of the Murdoch Children's Research Institute.

♦ Abby Lipman

♠ John Harris

in sociological terms what we would call the creation of an "ideal type". And the "ideal type" was genetic impairment or disabled people. That is to say in some of these writings you get a sense, (and it may be a shorthand and if you push them a bit they wouldn't really mean it, but it happens) - I am talking about the Nobel Prize winner, I am talking about a leading philosopher and I am talking about a lot of other people in this profession - a tendency to subsume all of genetic impairment and say it is all ghastly and terrible and has to be avoided. And if scientists and philosophers and clinicians can make this, what I call an "error" then is it any wonder that often in the media a similar error occurs. We have this sort of shorthand that genetic disease is terrible, we wish to do everything to avoid it and end of sentence. Now that rhetoric obviously makes those of us who have visible genetic diseases somewhat uncomfortable but I also don't think it is very helpful. It is not specific enough about what you and I think is important about the promise of genetic research and its clinical applications.

So, reductionism, geneticisation, the creation of an ideal type. And what I'm really here to say is that I think that there is a danger in getting carried away with the excitement of the human genome project, with the excitement of the molecular biology that you folk are doing, with the excitement of new diagnostic tests. And failing to see that for most of us, for many doctors in many ways it is business as usual…"

"So what I am beginning to get at here is that we need increasingly in the future a model where a genetic test or a genetic diagnosis does not mean that the solution is a prenatal termination. In some cases it may be, but in many cases it may be "Okay, let's design a society which is more accepting of disabled people." Let's design a education system which is able to cater to the needs of people with dyslexia or Down syndrome or whatever else it might be and enable them to reach their full potential. If it's breast cancer or colon cancer, let's have a screening service every year so we detect tumours and polyps and we can do something about that.

Quite clearly there is not a "one size fits all" model. Not every genetic disease demands a prenatal solution or indeed a genetic solution. I think we have to get our heads around that and of course all of you know that. That is a truism. But it's surprising how often people fail to observe that. Certainly in the media and I think often in professional discourse. As we find genes for more things - the behavioural genetic dispositions and all the rest of it - we are going to have to be more flexible in the way that we treat and react to genetic diagnosis. I am not saying that there is not a role for prenatal screening, I think of course there is and I think it is peoples right. I don't think it should be offered for everything to be tested. I think that clearly there are some pretty ghastly diseases and in my situation I would encourage a partner to have a termination in those cases. I'm not saying that we should ban genetics - of course not. I think we should have a choice. I think what you should be very careful about - the genetic profession - that society as a whole is saying that screening is the solution to genetic disease, and genetic disease is everybody and nobody wants to be genetically diseased because of course we are all genetically diseased."

The full transcript of Tom's presentation is available from the GSNV office.

Your Say…

In this section of the newsletter, you have the opportunity to write about your own experience of being faced with genetic challenges. Your letters and stories are always welcome.

Too big for a kids' hospital!

We are eagerly awaiting news of the opening of the adult clinic for people like my son Jason who has Glycogen Storage Disease Type 1a. At 34, he is getting just too big for the chairs and the Children's Hospital! Perhaps there could be an update on this in the GSNV News.

Gillian Davidson.

The Metabolic Services of Genetic Health have been aware for some time of the needs of adult patients to be treated in "adult" hospitals. We are negotiating with both the Royal Melbourne Hospital and the Monash Medical Centre to set up appropriate care for patients needing admission. In the meantime we are positioned to provide expert advice and management support to doctors in the event that any of our metabolic patients do need admission to an adult hospital. We will keep you posted of developments.

A/Prof Agnes Bankier
Director, Genetic Health Services Victoria
and
Dr Avihu Boneh
Director, Metabolic Service, Genetic Health Services Victoria

Media Scrapbook

Following are some highlights from newspapers in recent months. For full copies of the articles, please contact the Genetic Support Network of Victoria office on 8341-6315.

Gene research offers fresh hope

Researchers at Monash University are currently investigating the genes on chromosome 21 which are involved in Alzheimer's disease, leukaemia and heart failure in people who have Down syndrome. It is hoped that this work will benefit people with Down syndrome as well as other people affected by these three conditions.

A big smile but life can be tough going

The above story was accompanied by a story about, and great photo of, Rohan McDonald, son of Tony (secretary of the GSNV) and Arch. The McDonalds talked about how Down syndrome has affected their lives.

Alzeimer's cure coup

Herald Sun 22/12/00

Scientists report that they believe they may find a drug to cure Alzheimer's disease within five years, but the Alzheimer's Association of Australia warned against unrealistic optimism, and thought that 15-25 years might be a more realistic timeline.

Community grants online

Herald Sun 29/01/01

Rhonda Galbally (guest speaker at the GSNV AGM last year) reports on a new database for grants for small community groups. Check out www.ourcommunity.com.au for funding opportunities for your support group.

Scientists publish our genetic code

Herald Sun 12/02/01

The first draft of the human genome was published in the journal Nature in February. It is hoped that this information will lead to developments in development of drugs "customised" to the patient, as well as to earlier diagnosis of disease.

Genes may not be the answer

Herald Sun 13/02/01

Scientists report that the Human Genome Project indicates that humans have around

30,000 to 40,000 genes, which is far fewer than earlier estimates on 100,000. In fact we only have twice as many as a fruit fly!

Gene find to help diabetics

Herald Sun 8/02/01

Melbourne scientists at biotechnology company Autogen report discovering five genes involved in obesity and diabetes. These genes are part of a complex system of many genes and environmental factors which lead to these conditions.

Gene revolution begins

Herald Sun 17/02/01

This articles speculates on the potential impact of the Human Genome Project on medicine in the future, particularly with regards to drug development

Many articles about the Human Genome Project were printed in February. For more information on the Human Genome Project, visit the official site at http://www.ornl.gov/TechResources/Human_Genome/

GSNV Diary

Each newsletter we will publish a calendar of upcoming events such as, AGMs, seminars, conventions, social days, etc. If you or your group would like us to include details of your event please contact Caroline Bowditch at the Genetic Support Network of Victoria on Ph. (03) 8341 6315.

August			
Saturday 4th	**GSNV Annual General Meeting** *"What is the Human Genome Project? And what's it got to do with me?"* Guest Speaker Dr MaryAnne Aitken	10th Floor, Royal Children's Hospital	10am – 12 noon
Thursday 9th	**The Journey From Diagnosis** Week One of three-part seminar. All welcome – for enquiries call 8341-6315	10th Floor, Royal Children's Hospital	6.30pm – 8.30pm
Wednesday 15th	**Regular GSNV Meeting** All welcome!	10th Floor, Royal Children's Hospital	6pm
Thursday 16th	**The Journey From Diagnosis** Week Two of three-part seminar.	10th Floor, Royal Children's Hospital	6.30pm – 8.30pm
Friday 17th and Saturday 18th	**Australasian Leukodystrophy Conference** *"Leukodystrophy – treatment and cure: Working together with hope for a better future"* Tel: (03) 9584-7070 leuko@vicnet.net.au	Royal Children's Hospital	All day
Wednesday 22nd	**Public Forum with Dr Tom Shakespeare**	Melbourne Museum	7pm
Thursday 23rd	**The Journey From Diagnosis** Week Three of three-part seminar.	10th Floor, Royal Children's Hospital	6.30pm – 8.30pm
Sunday 26th	**Dialysis and Transplant Association (DATA) AGM** Tel: (03) 9894-0377	10th Floor, Royal Children's Hospital	12 noon – 6pm
September			
Sunday 2nd	**Short Statured People of Australia AGM**		
Tuesday 18th	**Decision-Making Processes for Families Affected by a Genetic Condition** Week One of two-part seminar series. All welcome – for enquiries call 8341-6315	MCRI 10th Floor, Royal Children's Hospital	6pm – 9pm
Wednesday 19th	**Regular GSNV Meeting**	10th Floor, Royal Children's Hospital	6pm
Tuesday 25th	**Decision-Making Processes for Families**	MCRI	6pm –

	Affected by a Genetic Condition Week Two of two-part seminar series.	10th Floor, Royal Children's Hospital	9pm