

QML PATHOLOGY

newsletter December 07

>>Genetic Test Advice - Principles and Example Cases - Part I

Dr Nigel Brown, Head of Molecular Pathology

The potential need to advise people considering having a 'gene test' is an increasingly common occurrence in all types of practice. While many types of genetic testing are not 'special', and require a similar level of advice and consent as any other pathology test, there is some genetic testing that goes beyond this.

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So the question arises “For this genetic test in this situation, what type of advice is needed and from whom?”

Answering the following questions will usually provide the answer:

- Why is the test being considered?
Is it for diagnosis or prediction?
- What are the implications of possible results?
- Does providing advice in this situation fall within my expertise?
- What other options for advice are there?

Answers to these questions will allow the practitioner to classify the testing into one of two levels recently defined by The Australian National Accreditation Advisory Council (NPAAC). NPAAC has categorised DNA testing into two levels as shown in Table 1 (reproduced from “Classification of Human Genetic Testing”, 2007).

These levels are:

Level 1 - requiring the usual degree of pre-test advice and consent.

Level 2 - requiring an additional level of pre-test counselling and consent.

Box 1 shows a series of criteria that arise from the definitions in Table 1. The laboratory will use these to guide classification of a test they provide as either a Level 1 or Level 2 test.

Laboratories are not required to sight details of the additional level of pre-test counselling and consent for Level 2 testing, but an indication that such informed consent has been obtained should be documented. This could be provided as an annotation on the request slip. Alternatively, it could be provided as an accompanying note or letter, perhaps from a Genetic Health Service if it was involved. Occasionally the requesting practitioner may be asked for further information on the advice and consent.

QML Pathologists can give advice on what level a test would be considered to be in a particular situation. Who would be most suited to give the additional level of pre-test counselling and consent can also be discussed if desired. This is a decision for the clinician based not only on the level of test but also on personal and professional aspects of the individual case.

^a The distinction between Level 1 (standard DNA test) and Level 2 (DNA test with potential complex issues) would usually be made by the doctor ordering the test, since that individual would be best placed to appreciate the short-term and long-term implications of the test for the patient and other family members.

Table 1: Levels of DNA Testing

Type of DNA test for an inherited genetic disorder	Explanatory notes ^a
Level 1 DNA test (standard)	Included here would be: a) DNA testing for diagnostic purposes (e.g. the patient has clinical indicators or a family history of an established inherited disorder, and DNA testing is being used to confirm the disorder) or any other DNA test that does not fall into Level 2. b) Neonatal screening programs.
Level 2 DNA test (i.e. the test has the potential to lead to complex clinical issues)	DNA testing for which specialised knowledge is needed for the DNA test to be requested, and for which professional genetic counselling should precede and accompany the test. Predictive or pre-symptomatic DNA testing, for conditions for which there is no simple treatment would usually be included in this grouping. Specific written consent and counselling issues are associated with this grouping.

Box 1: Schema for Classifying Human Genetics Tests

1. Genetic test requests for somatic variants are classified as Level 1 (e.g. testing for the BCR/ABL fusion gene in chronic myeloid leukaemia [Level 1]).

2. Genetic test requests for heritable variants, including diagnostic testing and medical screening programs, are classified as Level 1 testing unless a request fulfils one or more of the following criteria:

2.1. Guidelines developed by the National Health and Medical Research Council or a national medical specialty college recommend pre-test genetic counselling and written consent (e.g. testing for a familial BRCA1 mutation in a woman at high-risk of familial breast cancer [Level 2])

2.2. The specimen being tested is from a clinically affected child being tested for a disorder that typically presents in adulthood (e.g. testing for the Huntington disease mutation in a child with a neurodegenerative disorder [Level 2])

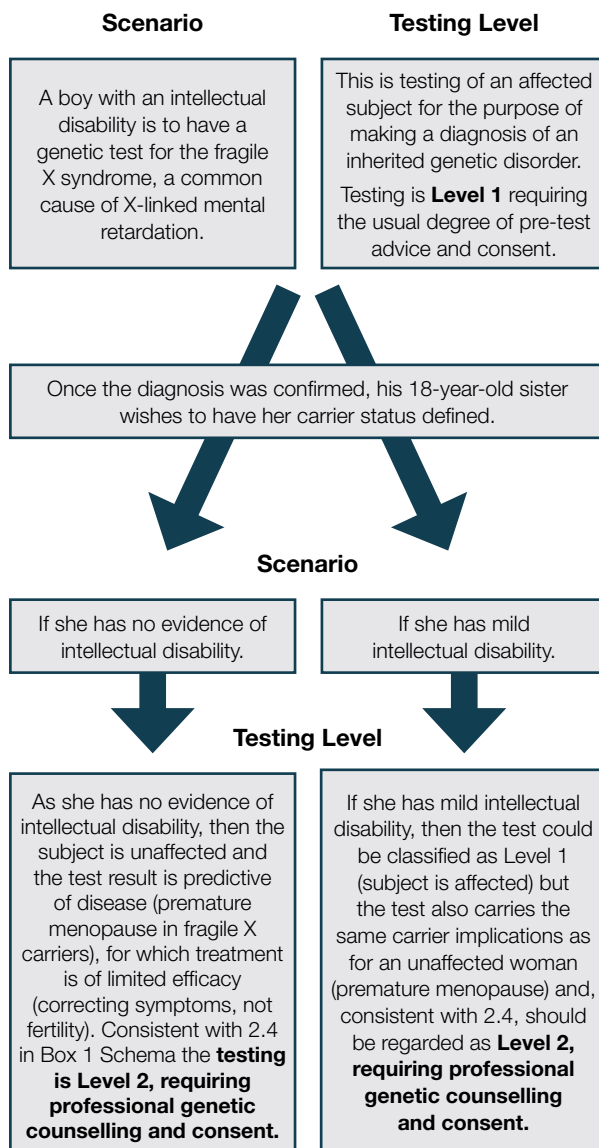
2.3. The specimen being tested is from an apparently unaffected child or foetus (e.g. prenatal testing for a mutation already defined in the family [Level 2]; carrier testing for Duchenne muscular dystrophy during childhood [Level 2])

2.4. The specimen for testing is from a clinically unaffected adult and the test is predictive of a disease for which interventions are of limited efficacy, or carry substantial risks or costs (e.g. pre-symptomatic testing for myotonic dystrophy [Level 2]).

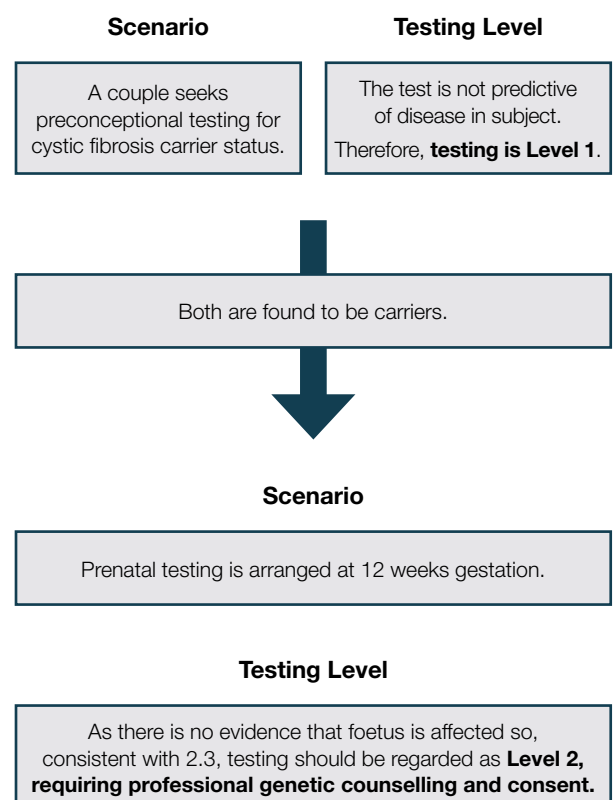
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Case 1



Case 2



Two cases, adapted from the document "Classification of Human Genetic Testing", 2007, are shown to give examples of how the particulars of a clinical scenario can determine different levels of genetic advice needed, even when the genetic 'test' is the same at the technical level. Case 1 involves Fragile X testing while Case 2 uses Cystic Fibrosis testing as an example.

A Part II of this series will further discuss available options for providing appropriate genetic advice.



Dr Nigel Brown
DipRACOG FRCPA

Pathologist in Charge -
Molecular Pathology &
Consultant Chemical Pathologist
Ph: (07) 3121 4428
Email: nigel.brown@qml.com.au

Dr Brown has been with QML Pathology since joining in May 1999 as Consultant Chemical Pathologist in the Biochemistry Department.

A graduate of The University of Queensland (1980), Dr Brown trained in pathology at the Royal Brisbane Hospital - obtaining his fellowship in chemical pathology in 1989. He remained at the Royal Brisbane Hospital for nearly a decade where, in addition to general chemical pathology, he explored his interests in genetics and errors of metabolism.

Special Interests: liver biochemistry, inborn errors of metabolism, computational biology, drug response genetics.

clinical data Dec 07

Infectious Diseases Report - Geographic Distribution - October 2007

SEROLOGY	Regions (as per key below)															Total			
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	Oct	Sep	Aug	Jul
Adenovirus (not typed)			1				2		3		2					8	23	54	20
Adenovirus (typing pending)																0	5	14	8
Barmah Forest virus		3		1			2			1	4				1	12	19	34	42
Bordetella pertussis			1				2		1	1	6	3				14	20	67	58
Brucella species																0	0	0	3
Campylobacter jejuni																0	0	0	0
Chlamydia pneumoniae																0	0	0	0
Chlamydia trachomatis, not typed	21	25	14	7			26		12	7	46	22	7	9	4	200	288	497	462
Coxiella burnetii	1			1		1	1		1			2	2	1	1	11	10	14	16
Cryptococcus species																0	3	2	2
Cytomegalovirus (CMV)	1	6	2				3		7	1	5	1	2	2	1	31	24	55	38
Entamoeba histolytica																0	0	1	0
Enterovirus - not typed		1							1			1				3	2	2	2
Epstein-Barr virus (EBV)	4	3	1				10		5	2	6	8	2	1	3	45	96	134	104
Flavivirus unspecified							1				2			1		4	7	23	10
Hepatitis A virus	1	1										2				4	1	0	5
Hepatitis B virus	2	2	2				3		1	2	9	1	1			23	47	75	80
Hepatitis C virus	5	13	4	3			9		9	2	25	7	4	2	3	86	135	252	204
Hepatitis D virus																0	0	0	0
Hepatitis E virus																0	0	0	0
Herpes simplex Type 1	5	14	3	1			9		6	2	23	5	1	2	3	74	120	221	272
Herpes simplex Type 2	5	15	3	1	1		17		6		18	9	1	3	2	81	105	180	177
Herpes simplex virus - not typed	1	4	1	1			3		1	1	7		1			20	14	47	67
HIV-1							1				1					2	1	1	4
HTLV-1																0	0	0	0
Influenza A virus		1					2		2	1		1	1		1	9	86	939	293
Influenza B virus																0	4	7	1
Legionella species																0	0	0	0
Leptospira species										1	1					2	2	3	2
Measles virus											1					1	1	1	2
Mumps virus																0	0	1	1
Mycoplasma pneumoniae	2	1	1				5	1	2		4	1		1		18	43	95	80
Neisseria gonorrhoeae	4	1					5				2	2		1	1	16	5	16	28
Parainfluenza virus Type 1																0	0	1	1
Parainfluenza virus Type 2																0	0	1	1
Parainfluenza virus Type 3			2				1		1	2	5		2	1		14	25	36	6
Parvovirus		2					3		3	1	3	2				14	24	47	28
Pneumocystis carinii		1														1	0	1	0
Respiratory Syncytial virus		4	1				7		4	3	8					27	55	130	83
Ross River virus		6	2			1	2		2	4	5	7	1			30	23	41	46
Rubella virus																0	0	0	1
Salmonella paratyphi A																0	0	0	0
Salmonella paratyphi B																0	0	0	0
Salmonella typhi																0	0	1	0
Shigella dysenteriae																0	0	0	0
Shigella flexneri																0	0	0	0
Streptococcus Group A	2	5	2		1		10	5	9	1	16	3	2	2		58	65	127	94
Toxoplasma gondii																0	1	0	1
Trponema pallidum	7	2	1		1	1	12		6	1	9	2		7	1	50	75	139	109
Trichomonas vaginalis	4		1											1		6	4	11	3
Varicella Zoster virus	7	7	6			1	9		5	3	9	6		2	4	59	116	175	181
Yersinia enterocolitica																0	1	1	0
TOTAL	72	117	48	15	3	4	145	6	87	36	217	85	27	36	25	923	1450	3446	2535

REGIONS

1 Cairns
2 Gold Coast/Northern Rivers
3 Ipswich

4 Mackay
5 Mount Isa
6 New England
7 North Brisbane Suburb

8 Northern Territory
9 Redcliffe
10 Rockhampton
11 South Brisbane Suburbs

12 Sunshine Coast
13 Toowoomba
14 Townsville
15 Wide Bay/Burnett

September 2007 and further historical clinical data can be obtained by contacting your local Medical Liaison Officer

This newsletter has been prepared and published by QML Pathology for the information of referring doctors. Although every effort has been made to ensure that the newsletter is free from error or omission, readers are advised that the newsletter is not a substitute for detailed professional advice.

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QML Pathology updates Dec 07

>> The Spirit of Giving

As the holiday season is upon us, we can reflect on the past year and on those who have supported us throughout this time. Historically QML Pathology has expressed our thanks with a small token of appreciation for members of the medical community. However, for the past few years we have been fortunate to have the opportunity to convey our thanks through a donation to assist those in the community who are less privileged than ourselves.

To give to people in need at this time is very gratifying and something each of you is a part of. It is through your continuing patronage and support that we have the opportunity to make a crucial difference in the lives of others. In 2007 Childhood Cancer Support and Corporate Angel Network of Australia will have the chance to convert \$50,000 into life-changing experiences.

Childhood Cancer Support

In response to the increasing incidence rate of childhood cancer in Australia, there is a growing need for services which provide support to these children and the family unit as a whole. Childhood Cancer Support (CCS) is a Queensland-based charity totally dedicated to the support and assistance of children with cancer, as well as their families.

The major aims of CCS are financial and emotional support, and the provision of accommodation for families affected by cancer. CCS owns and operates nine self-contained family units located close to the Royal Children's Hospital. Families stay free of charge in a 'home away from home' environment. In addition CCS provides recreational therapy activities, and continually advocates for childhood cancer research and improvements in treatment facilities.

For further information or to make a donation please call (07) 3252 4719 or visit www.ccs.org.au.

Corporate Angel Network of Australia

Long hours of travel by road or rail can be draining on a patient's strength, and in many cases makes visiting patients very difficult for families living long distances from treatment hospitals. Recognising that illness is traumatic enough, Corporate Angel Network of Australia aims to reduce the financial and emotional stress placed on these families.

Corporate Angel Network of Australia is a network of corporations and individuals in hospitals, cancer and leukaemia support groups, aviation companies and businesses who work together in order to provide free air transport to and from treatment centres for qualifying persons, namely cancer and leukaemia patients and bone

marrow donors and their carers anywhere within Australia. Their service supports families from Queensland and New South Wales, from the Northern Rivers region all the way up to Mt Isa.

For further information or to make a donation please visit www.angelnet.asn.au.

It is rewarding to have the opportunity to support those charities which give such wonderful financial and emotional support to your patients during some of the most difficult moments in their lives. Thank you to every member of the medical community, your support has enabled QML Pathology to help these charities make a difference in the lives others.



Graham Armstrong hands over our Christmas donation of \$25,000 to Helene McCarthy, Michael McCarthy and Dr Heather Parker, from the Corporate Angel Network of Australia.



Graham handing over the cheque for \$25,000 to Bob Kingston and Marg Ward of Childhood Cancer Support.

>> NEW Skin Pathology Collection Devices

At QML Pathology we are committed to providing the highest quality of service by embracing the latest technology and equipment. In addition to the recent introduction of the punch biopsy device with internal plunger, QML Pathology now has available BIOPBLADES™ for QML Pathology Skin Pathology referrers.

BIOPBLADE™

The sterile, single-use BIOPBLADE™ is a flexible scalpel used for cutaneous surgery, including: shave biopsy, saucerization of flat lesions and levelling of pedunculated lesions. The unique design of the BIOPBLADE™ incorporates a comfortable and protective 'Fingerguard' in addition to the flexible super sharp blade. This flexibility allows the blade to be positioned at the correct angle for the intended procedure.



The BIOPBLADE™ is utilized for removal of lesions, either elevated (shave biopsy) or flat (saucerization). After the site is anesthetized, the BIOPBLADE™ is held and 'bowed' between the thumb and fingers. The lesion is removed at or just below the surface epithelium. Cosmetic results are normally good and the wound heals without the need for suturing. The Clinician will remove all of the lesion without overly deep penetration to avoid scarring.



For further information or to order any of these devices, please contact our Stores Department on (07) 3121 4328 or your local QML Pathology Branch Laboratory.

Skin Punch Biopsy Devices

The punch biopsy with internal plunger system allows the lodged skin specimen inside the metal lumen of the punch to be easily ejected.

Type of biopsy	Sizes available (mm)
Punch Biopsy with internal plunger	2, 3, 4
Punch Biopsy without plunger	2, 3, 4, 5, 6, 8



Punch Biopsy with internal plunger



Punch Biopsy without plunger

Doctor's Noticeboard

- VERY large consulting room available for sessional use. Located inside QML Pathology's Robina Collection Centre the room is available any Monday, Wednesday or Thursday. The room is currently used by an ENT and Audiologist. Please contact Dylan on 0411 353 731 for further information.

WARFARIN DOSING OVER THE CHRISTMAS PERIOD

Over the upcoming Christmas period, the QML Pathology Warfarin Care Clinic will be closed. Please note that no new registrations will be taken from 6.00pm on Wednesday 19 December 2007, with the registration line re-opening at 7.00am on Wednesday 2 January 2008. During this period it is essential that any new patients on warfarin are supplied with instructions, as QML Pathology will be unable to monitor them until we re-open in January. Unfortunately QML Pathology cannot provide warfarin control for patients under a period of 4 weeks. As a result, we would appreciate if you could arrange a colleague to supervise any warfarin patients you control while you are on leave. Please note that this also applies to interstate patients on holiday in Queensland.