HOBART PATHOLOGY
LAUNCESTON PATHOLOGY
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Quality is in our DNA

Doctors' Newsletter

Issue 3 2018

Welcome	02
Specimen Collection from Specific Sites	02
Latent TB infection and cessation of the Mantoux test	02
Provision of Dexamethasone for testing	03
Know Pathology Know Healthcare	03
Anti -Mullerian Hormone	03
Reproductive Carrier Screening	04

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Welcome

This is the third Doctors' Newsletter for 2018 and, once again, it is my pleasure to introduce it.
The aim of the Newsletter is to provide articles that cover a broad range of topics relating to all the pathology sub-specialities with the aim to have clinical relevance and interest to the readership. In addition, the Newsletter is used to flag any changes to our policies that medical practitioners and their staff need to be made aware. In this current Newsletter, we have provided information on preconception testing offered through Sonic Genetics. We have also provided updated information on Mantoux testing, dexamethasone suppression testing and changes to Anti-Mullerian Hormone testing.
I hope you find the information presented in the Newsletters useful. If there are any articles that you would like to see in the future please contact our Client Services team or Dr Daniel Owens, the editor of the Newsletter. Thankyou for your continued support.

Dr Shaun Donovan
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CEO Diagnostic Services Pty Ltd (DSPL)
Hobart, Launceston and North West Pathology

Specimen collections from specific sites

Our Collection Staff have, on occasion, had requests to take the following samples:

-Skin scrapings for possible Scabies
-Vaginal swabs
-Urethral swabs
-Anal swabs

We are no longer able to offer collection from these sites.

Our Collection Staff can provide instructions and swabs to patients for self- collection of vaginal or anal swabs if requested.

Latent TB infection and cessation of the Mantoux test

We will no longer be offering the Mantoux test. Traditionally, the Mantoux test has been the investigation of choice in the diagnosis of Latent Tuberculosis Infection (LTBI).
The performance and reading of a Mantoux test requires the patient to present twice over a 2-3 day period for both administration of tuberculin and the subsequent subjective reading of the response.
Due to declining requests for the Mantoux test and the difficulty in maintaining expertise, the procedure will no longer be offered at our collection rooms. Please contact your local Respiratory Medicine Clinic* for advice about Mantoux testing.
We do offer Quantiferon® testing which is an interferon gamma release assay (IGRA) for LTBI diagnosis.
Quantiferon® has the advantages of being objective and requires only one attendance for the procedure to be completed.
In most situations this test attracts a Medicare Rebate. The current Medicare Rebate criteria for Quantiferon® testing are as shown:
Quantiferon® Testing : MBS Criteria (Item number 69471)

(a) a person who has been exposed to a confirmed case of active tuberculosis;
(b) a person who is infected with human immunodeficiency virus;
(c) a person who is to commence, or has commenced, tumour necrosis factor (TNF) inhibitor therapy;
(d) a person who is to commence, or has commenced, renal dialysis;
(e) a person with silicosis;
(f) a person who is, or is about to become, immunosuppressed because of a disease, or a medical treatment, not mentioned in paragraphs (a) to (e)

In situations where no Medicare rebate is available, the patient will be charged a non-rebatable fee of $75 to cover the cost of testing.
If there is a specific requirement for Mantoux testing, as opposed to Quantiferon®, the Respiratory Medicine Service* at each of the three major hospitals may be contacted as follows;

*Launceston General Hospital – Respiratory Medicine Unit
- Phone: 6777 6705
- Fax: 63487774
- Email: respiratoryunitlgh@ths.tas.gov.au

*Royal Hobart Hospital – Department of Respiratory Medicine
- Phone: 6166 7293
- E mail: margot.thompson@ths.tas.gov.au and/or rhh.respiratory@ths.tas.gov.au

*North West Regional Hospital – Respiratory Clinic
- Phone: 64936300
- Fax: 64306688

Provision of Dexamethasone for testing

Change to the provision of dexamethasone for the Low Dose Overnight Dexamethasone Suppression Test (DST)

The DST assists in the exclusion of Cushing's syndrome. Patients are required to take dexamethasone at 2300 the night prior to serum cortisol collection.
Our collection rooms will no longer be able to provide dexamethasone to patients.

When requesting a DST please provide your patient a prescription for dexamethasone 1mg (2 x 0.5mg).

Instructions to your patient:
Day 1 - take 1mg dexamethasone orally at 2300
Day 2 - present to a pathology collection room with request for DST between 0800-0900 for collection of a serum cortisol

Normal result: <50 nmol/L at 0900 post suppression.

For any further queries, please contact Dr Louise Prentice (Hobart Pathology) or Dr John Roberts (Launceston & North West Pathology).

Know Pathology Know Healthcare

know pathology
know healthcare

The Know Pathology Know Healthcare Initiative

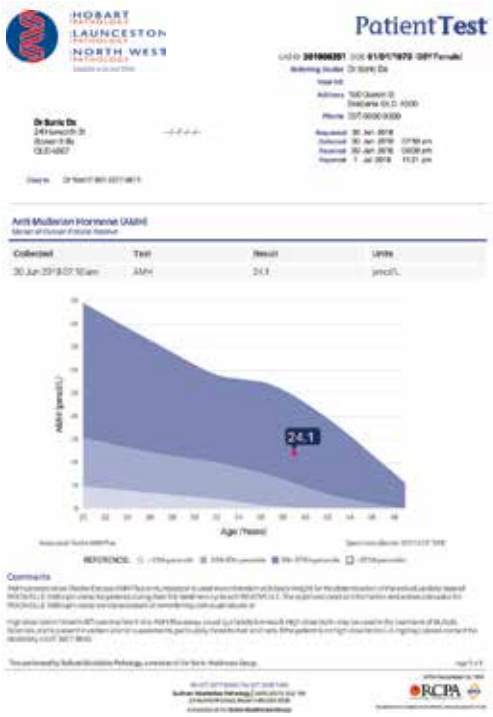
The Know Pathology Know Healthcare website provides patients with information on how pathology tests are performed, and how the results from those tests are used by doctors to diagnose and guide treatment. It also provides access to the Lab Tests Online database. This is a plain-English resource explaining pathology tests, curated by Australian Pathologists.

You can help us to improve patient health literacy by directing your patients to our websites if they would like to read more about their pathology tests.

A link to www.knowpathology.com.au is on our home pages.

Changes to Anti-Mullerian Hormone assay testing

As of September, we are changing our testing for Anti-Mullerian Hormone (AMH).
The new test system, the Roche AMH Plus assay, offers several analytical advantages, including the ability for the results to be used when dosing with the recombinant FSH drug, Rekovelle®.
Of note is that AMH levels are slightly different with the new assay, with the levels of AMH in the Roche assay running at 80% of the levels seen with the Beckman assay previously used. Conversion factor is as follows:
Roche AMH Plus Assay= 0.8 Beckman Access AMH Assay.
The Beckman Access AMH assay previously used will continue to be offered until 31st December 2018 to enable transition of patients who have had previous testing.
For information on the changes, please contact Dr Louise Prentice (Biochemistry Department) on 62371204.



What to order: AMH
Clinical notes: Record the day of cycle on request form and state if the patient is on contraception.

Sample :Serum tube, refrigerated transport.

Fee: No Medicare rebate. Private fee applies

Results will be reported as a text report with age-related reference intervals. As previously, a graphical report (above) will also be available electronically in PDF format or in hard copy as an additional report. It can also be viewed online using Sonic Dx.

Reproductive carrier screening

Professor Graeme Suthers: Director of Genetics Sonic Healthcare Australia

Single-gene disorders such as cystic fibrosis (CF), spinal muscular atrophy (SMA) and Fragile X syndrome (FXS) are more common than chromosome disorders in the children of younger mothers. Reproductive carrier screening can provide couples with information about the chance of such conditions and inform their reproductive choices.

A parent who is a carrier of a recessive mutation, that is, having one normal and one abnormal copy of a gene, will not be affected by the abnormal gene. Everyone is a carrier for one or more disorders; this is of no immediate consequence and there usually is no family history of the disorder. The situation changes if both parents are carriers of mutations in the same gene located on one of the autosomes (chromosomes 1-22). The chance of their child inheriting the abnormal gene from each parent, and so developing an autosomal recessive disorder, is 25%. The situation is similar for a woman with a recessive mutation on an X-chromosome: each of her sons is at 50% risk of inheriting the abnormal gene and being affected, and half of her daughters will be carriers. Overall, the risk of a woman who is an X-linked carrier having an affected child is approximately 25%.

There are hundreds of inherited recessive disorders that present in childhood. These disorders are individually rare but, together, they are more common than chromosome disorders. Further, the risk of these recessive disorders does not vary with maternal age. For mothers under 35 years of age, the risk of having a child with a serious childhood-onset recessive disorder is greater than the risk of having a child with a chromosome disorder.

Until recently, the only way of identifying a carrier of a recessive disorder was to diagnose the disorder in their affected child. This has now changed. It is possible to screen a couple for mutations in autosomal genes, and a woman for mutations in X-linked genes, to determine whether they are at 25% risk of having an affected child. This screening test is called 'reproductive carrier screening'.

Sonic Genetics offers reproductive carrier screening for CF and SMA (both autosomal recessive) and FXS (X-linked recessive). Approximately 6% of people are carriers of one or more of these conditions, and 0.6% (one in 160) couples are at 25% risk of having an affected child, Those couples who are identified as carriers can consider options, such as IVF with a donor gamete, pre-implantation genetic diagnosis, prenatal diagnosis by CVS, or they may make an informed decision to accept the risk. RANZCOG recommends that couples be offered information about such screening. The cost of this three-gene panel is approximately $400* per person. There is no Medicare rebate available; there are exceptions (and restrictions) for people with a documented family history of CF or FXS.

Sonic Genetics also offers the Beacon Expanded Carrier Screen of more than 300 genes which cause serious recessive childhood disorders. We estimate that approximately 70% of Australians are carriers for one or more conditions included in this screen and 3% (one in 30) couples are at 25% risk of having an affected child. This screen currently costs $995* per person or $1,750* for couples tested together.

It is important to consider some of the nuances in relation to Fragile X syndrome (carriers may develop adult-onset conditions), and when there is a family history of a recessive disorder (always seek expert advice). It is also important to recognise that some couples will not want this carrier information – and others will demand it. Each person needs to be free to make their own decision about what information they wish to have.

Sonic Genetics offers genetic counselling free-of-charge^ for couples who are identified by either of these screens as being at high risk of having an affected child. Further details regarding eligibility and process are provided on our website and with relevant test reports.

It is accepted practice that every woman is offered screening for chromosome disorders in pregnancy. In a similar vein, every couple should be offered reproductive carrier screening for recessive disorders, irrespective of age and family history. This simply represents good medical practice.

References

- RANZCOG. Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions. 2018 (August).
  Archibald AD, Smith MJ, Burgess T, et al. *Genet Med.* 2017; 20(5):513-523 [Available online doi:10.1038/gim.2017.134.]
- Sonic Genetics. (2018) Reproductive carrier screening [Online] <www.sonicgenetics.com.au/rcs>

*Prices correct at time of printing. ^Terms and conditions apply. Please refer to www.sonicgenetics.com.au/rcs/gc

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