virtus DIAGNOSTICS

WINTER 2018

NEWS

Welcome

In 2016 Virtus Health acquired my laboratory Independent Diagnostic Services, since then we have expanded our general pathology services under the Virtus Diagnostic brand throughout the eastern states of Australia. Today we offer a comprehensive range of testing across all pathology disciplines and have established world class genetic laboratories.

During the last 12 months we have successfully gained NATA corporate accreditation for all our laboratories, we have transitioned all of our general, genetics and andrology laboratories nationally over to a central Laboratory Information System (LIMS), expanded our collection centre foot print and recently launched our new website.

Our General Pathology laboratory has increased its scope of tests with the introduction of several new analysers in our chemistry and endocrinology departments. In October our new COAG analyser will be commissioned. With this new technology and the increase in testing capability we welcome new staff and scientists to Virtus.

Our Genetics laboratories are at the forefront of genetic testing in fertility and reproductive health. Our research and development ensures that our Genetic testing remains at world class standards. Over the next few weeks you will receive our new genetic information brochures and I look forward to your feedback and enquiries.

In this edition of our newsletter we outline two of our genetic tests;
Fragile X Syndrome (FXS) and **Spinal Muscular Atrophy (SMA).**

Both Fragile X Syndrome (FXS) & Spinal Muscular Atrophy (SMA) have been reported across all forms of media over the last few months. Our direct patient enquiries have increased as they make more informed choices about having these tests performed. Virtus Diagnostics offers both these tests as an individual test or as part of our Virtus Genetic Screen (VGS)

During my time as medical director we have seen rapid growth in our pathology business however we still pride ourselves in offering a personalised service to our referring clinicians. Myself, our pathologists and scientific staff welcome your enquiries and are always available to discuss any queries you may have concerning your patients or any of our services.

Thank you for your continued support.

Dr Tony Stoloff Medical Director

Specialist Genetic Testing

Virtus Diagnostics offers a specialist genetic testing service to all our referring clinicians.

For further information on our Specialist Genetic Testing log on to our website and click on the specialist-genetic-testing tab or phone our laboratory on

Ph: 1800 837 284

Our service offers:

- Specialists in clinical genetics
- A comprehensive range of tests
- Fast turnaround of results
- Highly skilled and experienced scientists, with access to the latest technology
- Convenient collection centres
- Personalised service

Fragile X Syndrome (FXS)

Fragile X Syndrome (FXS) occurs in approximately 1 in 4,000 males and 1 in 8,000 females. This X-linked genetic condition causes a range of developmental problems that encompass learning disabilities and cognitive impairment.

Males are usually more severely affected than females. The gene causing FXS (FMR1) is divided into three categories: normal, pre-mutation and full mutation depending on the repeat region length. The full mutation causes FXS. There is no cure for this disorder - educational and behavioural interventions may improve the outcome.

INHERITANCE: The syndrome is inherited from a carrier mother who has one X chromosome with the FMR1 gene repeat region in the pre-mutation range. The disorder is caused by an increase of the repeat region length to full mutation of the FMR1 gene. As males only have one X chromosome, when they inherit the full mutation they develop the disorder. Females have two X chromosomes and thus tend to have milder symptoms when they inherit the full mutation. With each pregnancy a carrier mother has a 50% chance of passing on the X chromosome with the pre-mutation repeat region. The chance it will expand to the full mutation increases with increasing size of the pre-mutation.

FRAGILE X AND PRIMARY OVARIAN INSUFFICIENCY (FXPOI) – PRE-MUTATION CARRIERS

Ovarian insufficiency occurs when the ovaries stop functioning normally in a woman younger than 40 years of age. Some women with POI can get pregnant, but once they have completed menopause they can no longer fall pregnant.

Studies of female pre-mutation carriers show that 25% experience ovarian insufficiency and 25% experience early menopause.

FRAGILE X AND TREMOR/ATAXIA (FXTAS) – PRE-MUTATION CARRIERS

This condition is characterised by problems with movement and thinking ability. It is a late-onset disorder (>50years) and the signs and symptoms worsen with age.

Symptoms include an intention tremor ie trembling or shaking of a limbs when trying to perform a voluntary movement and there are problems with coordination and balance. The tremors develops first and loss of balance may follow, but not all affected individuals exhibit both signs. There can be cognitive problems with loss of short term memory, the ability to plan and problem solve.

Male carriers of the pre-mutation occur with a frequency of one in 450 but only 40% of these will develop the condition. One in 200 females have the pre-mutation but only 16% develop the condition.

CASCADE SCREENING:

If you are shown to be a pre-mutation carrier and you have siblings and other close relatives, they may also be at risk of being carriers. Testing for these individuals is also available should they wish to determine their carrier status?

ACCURACY OF TESTING:

The test determines the number of triplet repeats in the FRX gene and determines if an individual falls into the normal, intermediate, pre-mutation of full mutation range. A small percentage of cases may require further testing.

THE FACTS:

- FXS is a major cause of inherited intellectual disability
- features range from mild to severe
- this is an X-linked disorder and males are more likely to be severely affected than females
- the disorder is inherited from pre-mutation carrier mothers
- presenting features in children include developmental delay, learning difficulties, anxiety, autism and hyperactive behaviour
- some individuals have seizures
- FX and primary ovarian insufficiency (FXPOI) – one in 50 women with an unknown cause of POI are Fragile X carriers
- FX and tremor/ataxia (FXTAS) is a late-onset disorder, usually after the age of 50 years and males who carry the pre-mutation repeat are more likely to be affected than females (40% vs 16%).

Spinal Muscular Atrophy (SMA)

Spinal Muscular Atrophy (SMA) is a neuromuscular disorder of the nerves. The nerves in the spinal cord are affected and the link between the brain and the muscles in the body is lost causing the muscles to become wasted. The progressive muscle wasting often leads to early death. The gene (SMN1) involved in this disorder has part of its genetic information deleted.

There are four types of SMA, with type 1 being the most common as well as the most severe. Affected individuals usually do not survive much beyond two years of age.

SPINRAZA® (nusinersen) injection 12 mg/5 mL

A drug, Spinraza, has recently been released and can be prescribed to help ameliorate the symptoms of the disease.

Usually there is no family history of the disorder as carriers are unaffected. Pre-conception screening offers couples the ability to determine their carrier status prior to pregnancy.

SMA is inherited in an autosomal recessive manner. For a child to have SMA **both parents must be carriers.** When two SMA carriers have a child there is:

- **a 1 in 4 chance their child will be affected by SMA**
- **a 1 in 2 chance their child will be a carrier of SMA but unaffected like the parents**
- **a 1 in 4 chance the child will not inherit the SMA gene variant and will not be at risk of passing SMA to future generations**
- **every pregnancy carries the same risk of conceiving an affected child as the previous one**

The gene for the disorder, SMN1 (survival of motor neuron protein) is found in all ethnic groups. The prevalence of affected individuals is estimated at 1 per 10,000 with one in 40-60 persons being carriers.

The majority of cases (approximately 95%) are caused by a deletion of genetic information in the SMN1 gene; however rare cases are caused by point mutations. In a small number of cases, neither parent is shown to be a carrier.

AN SMA SCREENING BLOOD TEST

is available at Virtus Diagnostics. If one member of the couple is tested initially and shown to be a carrier your doctor will recommend that your partner be tested. If both members of a couple are shown to be carriers, referral for genetic counselling can be arranged.

CASCADE CARRIER SCREENING

If you are shown to be a carrier, and you have siblings or other close relatives, they may also be at risk of being carriers. Testing for these individuals is also available should they wish to determine their carrier status.

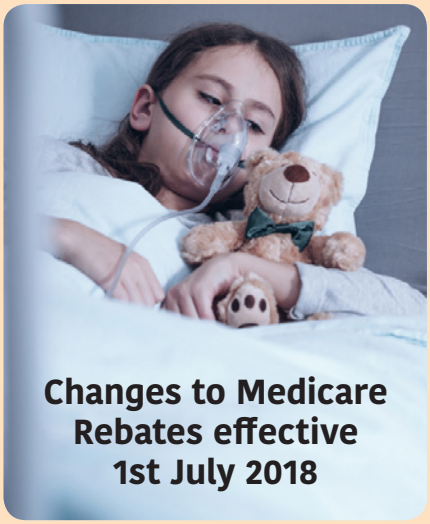
ACCURACY OF TESTING

This screening test detects the most common cause of SMA - 95% of all cases are due to a deletion in the gene.

THE FACTS:

1. SMA is a disease affecting nerves and the spinal cord resulting in muscles becoming progressively weaker
2. Carriers of SMA are normal
3. If both parents are SMA carriers the chance of having a child with SMA is 1 in 4
4. There are 4 distinct types of SMA
5. SMA type 1 is the most common and most severe
6. Survival for SMA type 1 is usually not greater than two years of age
7. Babies with SMA appear normal but are very weak
8. Children with SMA have normal intelligence
9. There is currently no cure for SMA

Cystic Fibrosis



Changes to Medicare Rebates effective 1st July 2018

As of 1st July 2018 new Medicare Item Numbers were introduced for screening for Cystic fibrosis (CF) carrier status for a defined set of individuals.

All requests for individuals to be screened where there is a known carrier in the family, MUST have a copy of the original laboratory report or full notation of the mutation detected. If this is not supplied then no Medicare rebate is applicable.

For further information on Medicare eligibility for this test please contact our accounts department on **1800 090 325.**

New Collection Centres Now Open

CASTLE HILL

62B Windsor Road
Castle Hill NSW 2145

MOUNT DRUITT

Pathology Suite
The Doctors Mt Druitt
Cnr Carlisle Avenue & Luxford Road
Mount Druitt NSW 2770

FOR OPENING TIMES
Phone 1800 837 284
or visit our website at
www.virtusdiagnostics.com.au

Virtus Diagnostics now has a NEW website!

Show-casing our Pathology and Specialist Genetics testing capability

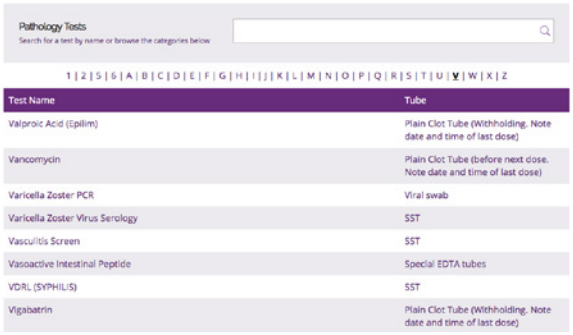
View your results ANYTIME ANYWHERE online

Visit us at virtusdiagnostics.com.au

Our updated site displays seamlessly, whether using your desktop, laptop, mobile or tablet, improving your experience with easy to navigate content which has been designed to enable fast, easy access to the information YOU need.

New Test Directory which includes:

- Alphabetical Search for pathology tests
- Tube/s required for collection
- Special instructions on each collection


Pathology Tests

Search for a test by name or browse the categories below

1 | 2 | 5 | 6 | A | B | C | D | E | F | G | H | I | J | K | L | M | N | O | P | Q | R | S | T | U | V | W | X | Z

Test Name	Tube
Valproic Acid (Epilim)	Plain Clot Tube (Withholding. Note date and time of last dose)
Vancomycin	Plain Clot Tube (before next dose. Note date and time of last dose)
Varicella Zoster PCR	Viral swab
Varicella Zoster Virus Serology	SST
Vasculitis Screen	SST
Vasoactive Intestinal Peptide	Special EDTA tubes
VDRL (SYPHILIS)	SST
Vigabatrin	Plain Clot Tube (Withholding. Note date and time of last dose)

Order pathology consumables online for your Medical Practice.

View your patient results anytime, anywhere through our web portal viewer.

Collection Centre Locations with easy to follow directions via a 'simple to use' interactive map.

Your patients can easily access Test Information and Test Instruction sheets for their procedures.

Secure, online payment facility.

We are updating our data base.

We are currently distributing forms requesting our referring clinicians to update their practice details, so that we may contact them in relation to critical results in a timely manner. All of this information is treated securely and in confidence and is only used in the case of extreme emergency. Thankyou for your participation.

LABORATORY LOCATIONS

SYDNEY - HURSTVILLE
12-14 Ormonde Parade
Hurstville NSW 2220

SYDNEY - GREENWICH
Level 3, 176 Pacific Highway
Greenwich NSW 2065

MELBOURNE
Level 2, 344 Victoria Parade
Melbourne VIC 3002

BRISBANE
Level 2, 55 Little Edward St
Spring Hill QLD 4000

For further information Phone 1800 837 284 or visit virtusdiagnostics.com.au