New test: Parathyroid hormone-related peptide (PTHrP)

Sullivan Nicolaides Pathology is now offering the measurement of parathyroid hormone-related peptide (PTHrP) for patients with non-PTH dependent hypercalcaemia. We are currently the only laboratory in Australia offering this assay with a 1–2 week turnaround time.

Hypercalcaemia of malignancy is a common complication of cancer and is due to the secretion of PTHrP by malignant cells. PTHrP is a protein existing in many isoforms with varying sizes. Both the N-terminus and the secondary structure of many PTHrP isoforms are able to bind to the same receptor as parathyroid hormone (PTH) due to their similar sequence and structure (see Figure 1). PTHrP can also be elevated in non-malignant conditions such as pregnancy, during lactation and in newborns.1

In malignancy, the binding of PTHrP stimulates calcium resorption from bone, renal reabsorption of calcium and osteolysis due to bony metastases. These events lead to malignancy-associated hypercalcaemia, driven by PTHrP secretion (see Figure 2).3,4 Patients with elevated PTHrP and hypercalcaemia have a poorer prognosis.5

What to request: PTHrP
- It is recommended that PTHrP requests are accompanied by a PTH and ionised calcium request.

Sample: 1 mL plasma
- (EDTA tube with Aprotinin/Leupeptin, available from Sullivan Nicolaides Pathology and included in the cost of the test).

Collection: Any SNP collection centre.
- Bookings required to arrange collection tube.

Transport: Sample is immediately separated and then frozen for storage and transport.

Cost: A Medicare rebate is not available for this test. A fee applies.

Turnaround time: The assay will be performed every 1 to 2 weeks.

References
Genetic testing for Familial Hypercholesterolaemia – patient brochure now available

Familial hypercholesterolaemia is one of the most common autosomal dominant disorders, affecting one in 250 Australians. With atherosclerosis starting in childhood, FH is a significant cause of premature coronary artery disease. However, at present the disorder is significantly underdiagnosed with an estimated 90% of Australians – or 90,000 people – going undiagnosed and under-treated.

Sullivan Nicolaides Pathology together with Sonic Genetics offers a comprehensive genetic testing service to assist specialists and general practitioners identify patients with FH. A diagnosis is important for the patient because:

- people with FH require earlier and more aggressive therapy to control their hypercholesterolaemia and reduce the risk of heart disease
- those with FH are eligible for selected medications on the PBS that may be necessary to achieve that control, and
- those who are identified as having FH and managed accordingly achieve better clinical outcomes than people with comparable cholesterol levels who are not identified as having FH.

FH is also important for the patient’s family because, in many cases, the condition is inherited as an autosomal dominant disorder, meaning that it is due to a single mutation in a gene. The children, siblings and parents of the index patient are at 50% risk of having the same mutation and developing hypercholesterolaemia. A genetic diagnosis of FH provides an opportunity to intervene and manage the condition before irreversible cardiovascular damage has occurred.

In order to support our referring doctors we have produced a plain language brochure that explains the nature and cause of FH, how genetic testing is used in diagnosis, and what a patient can expect during the testing process.

To order copies please contact your Medical Liaison Manager on 1300 767 284 or download from our website [https://snp.com.au/links/fh/](https://snp.com.au/links/fh/).

Help your patients understand their reproductive carrier screening options

RANZCOG now recommends that information about reproductive carrier screening for common disorders be offered to every woman either prior to conception (preferred) or in early pregnancy.

Sullivan Nicolaides Pathology, together with Sonic Genetics, offers two types of reproductive carrier screening – a three gene panel and a comprehensive >400 gene panel.

To support our referring doctors in helping their patients make an informed choice, we have summarised the key points for each test in an information sheet.

Download the information sheet from our [https://snp.com.au/links/carrierscreen](https://snp.com.au/links/carrierscreen) or contact your Medical Liaison Manager on 1300 767 284 to order copies.


Be prepared for Christmas

Warfarin Care enrolments

To ensure the safe and complete enrolment of patients into our Warfarin Care program, enrolments will be closed between the following dates:

**Community patients:** closing 5 pm Friday 4 December 2020 and re-opening Monday 4 January 2021.

**Hospital patients:** closing 5 pm Wednesday 9 December 2020 and re-opening Monday 4 January 2021.

**Stores**

To avoid running out of essential stocks over Christmas, such as personalised request form books and consumables, we advise our referrers to order stores before Friday 11 December for delivery before 23 December. Our stores department will be open to accept orders over the Christmas period, with despatch to recommence on December 30.

Collection centres

Please see [www.snp.com.au](http://www.snp.com.au) for updates to pathology collection centre hours of operation.

Cardiology services

Cardiology services will be available up until Friday 18 December with all results issued by COB Wednesday 23 December. Appointments will recommence from Monday 4 January 2021.

The Cardiology Department will be closed from 4:30 pm on 23 December and re-open on Monday 4 January 2021. For access to results during this time, please contact Dr Services on 1300 778 555.