JUNE 2016 | Published by RCPA

Issue #059

In This Issue

Interesting Facts

130,466

The estimated number of people who will be diagnosed with cancer in Australia in 2016.

100%

The proportion of these cancer diagnoses that will be made by a pathologist.

Source: RCPA Facebook page

Welcome to the June edition of ePathWay

It's likely that few people outside of medicine have heard of Raynaud's disease, Lynch Syndrome and subacute sclerosing panencephalitis (SSPE) – and that's why we've covered them. Their impact deserves a higher profile.

In contrast, most people have probably heard of altitude sickness but may be surprised to find out that its causes are still being debated. We found out why.

Don't forget to check our posts on our Facebook page (we've got over 1,300 likes). You can also follow our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter to keep up to date with pathology news.

Deadly measles-related brain infection targets children

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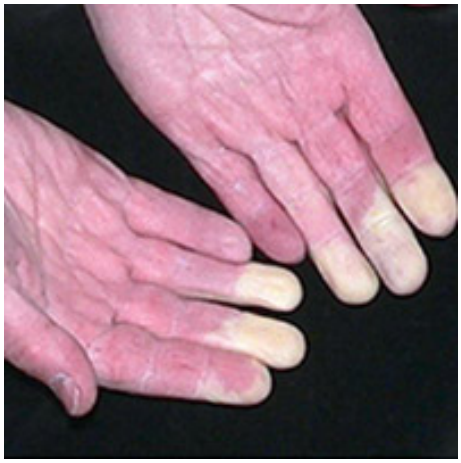
When most people think of winter ailments, coughs, colds and the flu probably come to mind. Raynaud's disease probably doesn't get a look in – unless you suffer from it.



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Deadly measles-related brain infection targets children

Subacute sclerosing panencephalitis (SSPE) is as bad as it sounds. It's a progressive, debilitating brain disorder associated with measles infection that typically affects children, and results in death.

Associate Professor Renate Kalnins, Anatomical Pathologist at Austin Health in Melbourne, says SSPE can affect anyone, but it mostly occurs in children who had developed measles before the age of two years. Symptoms appear years later, usually when the child is between five and 15 years old.

"It's thought that the measles virus mutates and persists in the brain's nerve cells because the viral particles are not able to be cleared away by the immune system. This eventually leads to degeneration of brain tissue and death of nerve cells, and these changes can spread to all parts of the brain, the brain stem and spinal cord," she explains.

Symptoms include subtle behavioural changes that progress over one to three years to fine motor problems, dementia, blindness and death, usually from pneumonia.

Dr Kalnins says the risk of contracting SSPE is not inconsequential. Cases have been reported at a rate as high as one in 4,635 naturally acquired measles infections in the USA, and one in 1,700 to one in 3,300 for children in Germany.

"The diagnosis of SSPE can be confirmed by a brain biopsy to look for characteristic features such as inflammation, gliosis (scarring), fewer neurones and aggregates of measles viral particles in cells. If these are not visible, PCR testing can be used to see if the virus is still present."

Dr Kalnins says another brain disorder that can occur secondary to measles infection is acute disseminated encephalomyelitis (ADEM). This results in a usually self-limited inflammation in the brain and spinal cord that damages myelin (the protective covering of nerve fibres).

"ADEM after measles infection affects about one in 1,000 people, but this rate drops substantially to one in two million

amongst people vaccinated for measles," she explains.

"SSPE, and to a lesser extent ADEM, are certainly devastating consequences of a measles infection, especially since SSPE is almost always fatal and affects mostly children. As a society we need the herd immunity bestowed by vaccination to keep the highly contagious measles virus in check."

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Scaling the world's highest mountains comes with many inherent risks, including altitude sickness. This is not a single ailment but a group of illnesses that develop at high altitudes, and the symptoms mostly reflect the body's response to insufficient oxygen supply. We asked RCPA Fellow Professor David Murdoch, co-author of *The High Altitude Medicine Handbook*, to walk us through this condition.

Prof Murdoch, who has lived in the Everest region, says there are still debates about how and why altitude sickness happens.

"This probably reflects the remote locations where this illness occurs, and the fact that there aren't any substantial medical facilities on hand in these areas, including pathology laboratories, when the illness is in the acute phase. It's also because of these remote locations that caution is needed if a person is thinking of climbing to a high altitude and they have existing medical issues," he explains.

For example, Prof Murdoch says people with chronic lung conditions who have medical issues at sea level will also have issues at altitude.

"People with heart disease also need to be cautious, because while age is not a big issue, exertion on the body is an issue when it comes to altitude sickness. But there are lots of different potential risk factors. The most useful one is a previous history or episode of altitude sickness, but this is not very helpful when you are on the mountain for the first time."

Prof Murdoch says the most common form of altitude sickness is Acute Mountain Sickness (AMS) which can kick in at about 2,000-2,500 metres. Its symptoms include headache, insomnia, tiredness and mild shortness of breath.

"The causes and effects of AMS are still being debated, but one theory is that it is possibly a mild form of cerebral oedema (swelling around the brain). About 50 per cent of people who trek to Everest Base Camp will develop AMS, and for most people it goes away once they rest and they won't have any further complications."

The most dangerous forms of altitude sickness are High Altitude Cerebral Oedema (HACE) and High Altitude Pulmonary Oedema (HAPE) in which fluid collects around the brain or lungs respectively. Symptoms of HACE include confusion and loss of coordination (ataxia), and it can result in coma or death. HAPE symptoms include shortness of breath, a cough and blood stained sputum. It's the most common cause of fatal altitude sickness.

"Altitude sickness also creeps up on you. It might start with a headache and then the symptoms can gradually become worse. Other physical changes might include fluid retention leading to swelling of the face, hands and feet - and we're not sure why - and a change in the pH of the blood. It's also risky sleeping at altitude because our breathing changes and slows down when we're asleep."

Amazingly, the human body will try to adapt to altitude, but it needs time. Prof Murdoch says after about a month or longer at altitude the blood becomes thicker due to more red blood cells being produced to carry the limited supply of oxygen around the body.

There is no medical test for any form of altitude sickness. Diagnosis is based on clinical signs and symptoms. When a person dies at altitude, Prof Murdoch says the post mortem might show swelling around the brain and lungs, and fluid retention, and other causes of death are usually ruled out first.

Despite its toll on mountaineers, altitude sickness seems to be one of those conditions where the more we find out about it, the more we realise we don't know about it. The risk of developing it also comes with the territory, and that territory is often far removed from comprehensive medical and testing facilities.

Prof David Murdoch is Head of Pathology at the University of Otago in New Zealand, and a clinical microbiologist. He co-authored *The High Altitude Medicine Handbook* with Andrew Pollard.

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Lynch Syndrome is responsible for about five per cent of all colorectal cancers, but it has a surprising low profile. It is caused by an inherited gene mutation that significantly increases the lifetime risk of developing not only colorectal cancers, but other cancers as well.

Dr Robyn Laurie, Anatomical Pathologist at Dorevitch Pathology in Melbourne, says the genes affected by Lynch Syndrome are responsible for repairing mistakes that occur in our DNA during cell division.

"People with Lynch Syndrome develop premalignant colorectal polyps at the same incidence as other people, but they are born with a mutation in one of their mismatch repair genes. If the second copy of that gene mutates in a polyp, then the lesion can progress to cancer at a faster rate compared to a person who doesn't have this syndrome," she says.

Dr Laurie says apart from colorectal cancer, people with Lynch Syndrome also have an increased risk of developing non-gastrointestinal cancers such as endometrial and ovarian cancer, and cancers of the pancreas, small intestine, stomach, renal pelvis and ureter.

“It’s very important to recognise Lynch Syndrome cancers because the genetic link puts the genetic relatives of the person diagnosed with it in the firing line as well. This will also mean adjusting screening and monitoring regimes to ensure potential cancers are caught early. Treatment regimes for Lynch Syndrome cancers can also be different to non-Lynch Syndrome cancers because they will often respond to different chemotherapy drugs.”

Diagnosis of a Lynch Syndrome cancer requires the expertise of a pathologist who picks up clues when they are looking down the microscope at cancer tissue.

“It might be the morphology (form and structure) of the tumour or the presence of lymphocytes in a tumour that indicate it's a

Lynch Syndrome cancer. If we suspect it is, then we will perform special stains on the tumour to confirm the diagnosis. The diagnosis can then be confirmed by molecular testing," explains Dr Laurie.

“There are also sub-sets of the syndrome, such as Muir Torre Syndrome, to be aware of as well. This syndrome results in colorectal cancer and sebaceous (skin) tumours occurring together,” she says.

The good news is that people with Lynch Syndrome cancers often have better survival and recurrence rates if the cancers are caught early. The key is identifying these cancers, and then ensuring genetic relatives are also screened for the genetic mutation, to help prevent or catch all potential Lynch Syndrome cancers early.

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When most people think of winter ailments, coughs, colds and the flu probably come to mind. Raynaud's disease probably doesn't get a look in – unless you suffer from it.

Raynaud's disease is a rare disorder of the blood vessels that causes them to narrow and spasm in response to cold or stress. It commonly affects fingers and toes, but can also affect other areas such as the nose, lips, nipples and ears.

During an attack, the affected areas of skin typically turn white before they feel cold and numb and turn blue. When the affected areas are warmed again they may turn red, and throb, tingle or swell. Most people have no long-term tissue damage.

Primary Raynaud's is the most common form, and while its cause is unknown, it isn't the result of an underlying medical condition. That's the territory of secondary Raynaud's which is the more serious form of this disease, and can be caused by injuries, certain medicines and other diseases.

"When a person presents with symptoms of Raynaud's disease, ruling out underlying diseases, including significant autoimmune conditions, is very important," explains Dr Richard Steele, Clinical Immunologist and Immunopathologist in Wellington, New Zealand.

"For this reason we will run autoimmune serology blood tests and especially rule out systemic sclerosis as the underlying cause. This is because this disease is a common cause of secondary Raynaud's and can be very devastating."

Systemic sclerosis results in abnormal growth of connective tissue causing damage to blood vessels and thickening and scarring of connective tissue (fibrosis) in the skin, gastrointestinal tract and other internal organs. Dr Steele says other autoimmune diseases associated with secondary Raynaud's include rheumatoid arthritis and systemic lupus erythematosus (SLE).

"Raynaud's disease is also more common in women than in men and we don't know why, but we do know that autoimmune diseases typically occur more frequently in women," he explains.

Dr Steele says while there is no specific test for Raynaud's disease, and diagnosis is based on signs and symptoms, an antinuclear antibody (ANA) blood test is useful to check for autoimmune disorders that affect tissues and organs. He says if this test is positive, then a further test looking for antibodies associated with systemic sclerosis is often the next step.

While Raynaud's disease is typically associated with colder temperatures, people living in warmer regions are not in the clear. It can occur in warmer climates because even mild or brief changes in temperature can trigger attacks, as can taking frozen foods out of the freezer with bare hands.

Raynaud's isn't a common winter ailment, but it would be alarming when it first strikes. The important next step is ruling out any nasty underlying causes, especially systemic sclerosis, to make sure it's is not a white, blue and red warning sign that something else is amiss.

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