EMERGENCY MEDICINE ONE STEP CLOSER

Professor Ralph Kirsch and Dr Clive Balfour.

Emergency Medicine (EM) last month reached a milestone on the tortuous road to registration as a full specialty with approval by the Education and Registration Management Committee of the Medical and Dental Professions Board (MDPB).

The chair of the Postgraduate Education and Training Committee, Professor JP van Niekerk, confirmed to the SAMJ that both committees had recommended to the MDPB that EM registration be approved.

However, at an earlier briefing of interested professionals hosted by the honorary head of UCT's new EM division, Dr Clive Balfour, Van Niekerk was quick to temper expectations and warned about the slowness of bureaucratic machinery. He said he would be 'surprised if registration came within two years'.

He told the SAMJ that he had asked for it to be put on the agenda of the next meeting of the full MDPB on 3 March. If it missed that cut, the 'Recommendation for Approval' would have to languish until the second annual Board meeting in September.

From there, the Ministry of Health would put out the proposed registration for comment for three months before the specialty was gazetted and came into being.

Van Niekerk reminded attendants that occupational medicine's application for registration had been awaiting publication at the Department for two years, adding that 'sometimes there are just structural inefficiencies of which we need to be aware'.

He added that the Minister had representatives on the MDPB who could theoretically 'fast-track' the application, but the practice did not always match the theory.

Opening the briefing session, Professor Nicky Padayachee, Dean of the Faculty of Health Sciences at UCT, said it had been 'music to his ears' when he first heard Balfour's proposal for a Chair of EM at UCT.

In the Western Cape, 70 000 tertiary hospital admissions per month are attended to by 200 doctors, less than 10% of whom have done all three Advanced Life Support Courses.

'The night before, I had spent four hours with the Minister who said the biggest problem in southern Africa was trauma. Early intervention can make a remarkable difference in sequelae.' Her sentiments were supported by another influential Cabinet member, Transport Minister Dullah Omar, who was excited when told about the latest EM moves towards registration.

Padayachee added that 'even in sophisticated countries like Germany, trauma is still a major discipline'.

He said he had encountered resistance from UCT colleagues who believed EM would be too expensive and steal resources. They had urged him to 'send it to Tygerberg if you can'.

However, Padayachee nailed his colours firmly to the mast, saying a spread of EM training to Pretoria and Wits campuses would dramatically reduce hospital bed occupancy.

'If we can market and promote it, I don't think we'll run short of resources,' he added, promising cross-disciplinary support in the hunt for resources.

Professor Ralph Kirsch, UCT Dean of Medicine and President of the Colleges of Medicine, was upbeat about the chances for EM registration before the end of this year.

'I can understand Professor Van Niekerk's reticence but President Thabo Mbeki says trauma is the major cause of death in South Africa and I'm sure he'll support it,' he said.

Kirsch said the EM program at UCT was 'already generating income' and emphasised 'the need to be self-sufficient in a non-specialist environment'.

He said an underpinning philosophy and enabling environment existed in the Western Cape and he promised to donate some of his Medicine Department posts to the EM programme. 'We're quite prepared to donate half our MO posts,' he said.

The Colleges of Medicine had obtained agreement from the HPCSA to form a College of Emergency Medicine and fellowships would be awarded by peer review.

In a fact-filled presentation garnered from his years of researching the subject internationally, Balfour said trauma formed 25% of admissions in South Africa compared to 8% in the USA and 6% in the UK. Africa was more in need of emergency medicine doctors than any other continent.

Illustrating the need for EM in the Western Cape, he painted 'a terrifying picture' of 70 000 tertiary hospital

admissions per month being attended to by 200 doctors, less than 10% of whom have done all three Advanced Life Support Courses. 'There is simply no minimum training requirement.'

Balfour revealed that it took from the October 2000 inaugural meeting of the SA EM Society to December 2002 to get the matter before the HPCSA.

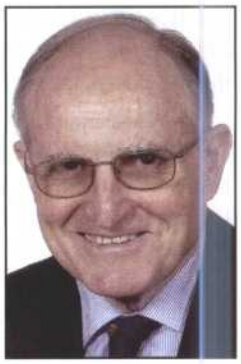
UCT was the first of South Africa's eight medical schools to offer EM. The Universities of Pretoria and Wits were developing EM programmes, none of which were yet running, but all of which would eventually collaborate with UCT.

Outlining UCT's plans, Balfour said he hoped to have secured a four to eight week EM block in the final undergraduate year by next year, set up a generic life-support course and a clinical skills laboratory.

Higher degrees in EM included an M-Phil (the first students were busy with dissertations and had written the exam), a Master of Medicine (once the specialty is registered), and Fellowship of the Colleges of Medicine.

Exchange programmes were being lined up with Yale University and the London Institute of Technology, while fellowships would be available at Yale and Edinburgh universities.

Asked about the 'Grandfather Clause,' Balfour said a table would have to be developed based on international standards to 'prevent any unnecessary heartaches and mistakes'.



Professor J P van Niekerk.

He believed a credentials committee should be formed with fellowship as the gold standard, while a points system would be the fairest means of assessment.

Chris Bateman

LITTLE KNOWN KILLER: BARTH SYNDROME

Gene carriers Jeanette and Alison with sons Benjamin and Colin.

Two KwaZulu-Natal sisters whose sons are South Africa's only known sufferers of Barth syndrome, have spoken out in an attempt to save the lives of undiagnosed boys.

Jeannette Thorpe has become a lay expert on this treatable but incurable genetic disorder carried by mothers and which affects only sons. 'I can't believe there are only two of us in the country. I'm terrified there are children out there dying of this disease through incorrect diagnosis - we want to make the disorder known'.

The former human resources consultant from Kloof returned to work soon after the birth of her son Benjamin and travelled with him around the country during his first year.

'He was carried full term, weighed 2.4 kg at birth and weight gain for the first six weeks was fairly slow. From eight weeks he began losing weight, looked uncomfortable, cried and sweated when breastfed. He became hysterical if I laid him flat,' she told the SAMJ.

Her paediatrician discovered that Benjamin was in cardiac failure and immediately referred him to a paediatric cardiologist. On examination, he was recessing quite markedly and was tachypnoeic. There were no dysmorphic features. His liver was 2 cm below the right costal margin and was firm.

Heart sounds were normal with no murmurs but there was a soft gallop present. An echocardiogram showed a dilated, hypertrophied, poorly contracting left ventricle. Benjamin had a severe cardiomyopathy.

In order to exclude an inborn error of metabolism as a cause, the paediatric cardiologist ran various tests. These came back normal except for slightly raised lactate. Urine and serum organic acids were sent to a Potchefstroom laboratory and came up normal except for a slightly raised 3-methylglutaconic acid. Carnitine levels were also normal.

The opinion was that this metabolic profile was 'acceptably normal'. The reason for the slightly raised 3-methylglutaconic acid and lactate was 'unknown' but as a single entity this was 'probably not indicative of a metabolic disorder'.

Two years later, Jeannette's sister, Alison Campbell-Gillies, gave birth to Colin seven weeks prematurely. Colin also failed to thrive and at six weeks was in cardiac failure.

A second paediatric cardiologist examined both children and found virtually identical symptoms - neutropenia, muscular weakness and cardiomyopathy.

After a lengthy search he found an article in the Journal of Neurological Science that described an x-linked cardioskeletal myopathy known as Barth syndrome.

The syndrome was first described in 1981 by Dr Peter Barth of the Netherlands who had been following a

family with an extensive history of male infant mortality. Despite efforts to save these children, they all died.

Upon hearing of the grave condition of the last affected child in the Dutch family, Barth asked that the family bring the boy into hospital. The state of his muscle tissue revealed in the later autopsy started Barth on research that ultimately led him to identify the condition and its genetic cause.

Prior to the publication about the Dutch family in 1983, only one family with a similar, possibly x-linked disorder had been described in the medical literature by Neustein in 1979.

In the 10 years following Barth's 1983 report, at least 25 additional cases were described with the same triad of cardioskeletal myopathy, neutropenia and growth retardation.

Maternal carriers show no symptoms of the disorder but there is a 50% chance that any sons will suffer from it, while girls born to a carrier have a 50% risk of being carriers themselves. All daughters of a male with Barth syndrome will be carriers, but their sons will be clear.

Now eight years old, South Africa's Benjamin Thorpe still has severe cardiomyopathy with poor function and slight neutropenia. His cousin, six-year-old Colin, lives with chronic neutropenia and a myopathic heart with good function. He receives a weekly injection of Neupogen.

Two years ago both boys went into cardiac failure due to infections, but have since been stabilised.

The boys have the physique of children half their age which is common among Barth patients. Colin walks with the typical waddling gait. They tire easily and have to temper their physical exertions and are medicated three times per day. Growth retardation becomes largely resolved after puberty - one of the issues that makes Barth syndrome stand out in contrast to all other mitochondrial syndromes.

Benjamin and Colin are academically bright, outspoken and sociable children. Colin's passion is playing tennis and Benjamin is a 'water baby'. The boys seem to know when they have had enough action, but occasionally their mothers must step in to slow them down.

Since they are accustomed to regular blood tests in hospital, Benjamin and Colin have since decided that they would both like to become doctors.

Jeannette is so fired up about creating awareness because 'an early diagnosis means a child has an 85% to 90% chance of survival. Those not accurately diagnosed have a 30% chance of living through the first few years of life'.

Experts advise doctors to be highly suspicious of any history of infantile deaths or underweight births and subsequent weight loss.

The sisters are part of a global group of physicians and families called the Barth Syndrome Foundation, a non-profit organisation with its own top-grade scientific advisory board.

The board is chaired by Dr Richard Kelley, the Director of Metabolism at the Kennedy Krieger Institute and an Associate Professor of Paediatrics at Johns Hopkins University.

Members of this team span related clinical specialities such as cardiology, neurology and haematology, as well as areas of research expertise such as biochemistry, molecular genetics and lipid metabolism.

The vision of the Barth Syndrome Foundation is 'a world in which not one more child will suffer or perish from this condition'. Its mission is to guide the search for a cure, to educate and support physicians and to create a caring community for affected families. Their website is at www.barthsyndrome.org.

The experts advise doctors to be highly suspicious of any history of infantile deaths or underweight births and subsequent weight loss. (Alison lost a baby boy in 1985 - the cause at the time was unknown.)

While genetic testing is available for Barth syndrome in Holland and America (the disease gene is called G4.5), a laboratory in Potchefstroom can run simple urine tests for raised 3-methylglutaconic acid.

According to Kelley, raised levels of this acid appear to be largely independent of the severity of the other features of the disorder. 'Levels are especially high between the ages of six months and three years and can be up to 200 times normal.'

He added that there are no good studies of the population or birth incidence of Barth syndrome. However, probably fewer than ten new Barth infants are identified each year in the United States, which suggests an incidence of only 1 in 300-400 000 births and a true birth incidence of probably no more than 1 in 200 000.

The long-held suspicions of metabolicists and geneticists in Bristol, England, have been proved right, i.e. that Barth syndrome is more common than generally recognised, perhaps because affected children present to so many different medical specialists.

In the past 20 years in the United States, only 10% of diagnosed Barth syndrome patients have died, whereas 70% of their older siblings had died before the syndrome was recognised.

Similarly, once neutropenia is recognised, its complications are largely preventable by close monitoring of the patients and prompt use of antibiotics.

Before the formation of the Barth Syndrome Foundation, Jeannette and Alison said it was 'terrifying' to face the battle alone. 'We had no idea what we were facing. There is now more knowledge. Never again should any family feel so desperately isolated and lost'. They are determined to spare other parents their ordeal.

Chris Bateman