

'My patient's strong desire to have children led her to The London Clinic from Kuwait, in search of another expert opinion.'

Mr Colin Davis page 3



theNotes

One Gene or Two?

Clinical Genetics within Modern Medicine Today

Ultimately medical genetics is about the patient and is trying to take us from the gene to the molecule to the organ and how that fits in with the body in general. Therein lies the continuing hope for the future.



The London Clinic
Newsletter
Summer 2006

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When I tell people I am a clinical genetics doctor they say, "Oh, that's a shame, do you miss not seeing the patients now?" Sadly this is how many people view genetics and the images portrayed by the media and artists like Jake and Dinos Chapman in the recent exhibition at the Saatchi Gallery help perpetuate this idea of genetic monsters.

Personally I see patients all the time, working closely with Cytogenetic and Molecular laboratories. Medical Genetics is a diagnostic and counselling service for individuals or families concerned about the suspected or confirmed diagnosis of a genetic disorder. This may include affected individuals or those concerned about their risk on the basis of their family history. Other common reasons for referrals are families with a history of reproductive loss or congenital abnormality and developmental or sensory disability.

Since Langdon Down first described Down Syndrome in 1865, we have learnt to recognise many more typical facial appearances across a variety of disorders. We describe new syndromes even now. Our understanding of genetics inheritance and DNA structure has evolved tremendously in a short time given that chromosomes were first described in the 1940's. We are now able to look at the substructure of chromosomes, in particular the tips of the chromosomes called telomeres.

Mutations in these ends can be the cause of developmental delay in up to 8% of previously undiagnosed cases. We can sequence genes and identify small point mutations. However it remains a clinical diagnostic speciality since we need to know which disease gene to test.

Clinical Genetics is a service which offers clinical diagnosis, laboratory investigations, risk estimation and explanation of this to the family to enable them to decide on the most appropriate course of action and to understand how technology may benefit them. Establishing a genetic diagnosis or risk may also help colleagues in primary and secondary care to choose the most appropriate patterns of treatment and care.

Medical Genetics used to be mostly about trying to diagnose children and the various conditions they had but increasingly now we are looking at the genetics of adult conditions and other common later onset disorders. Family histories of cancer and in particular breast and or bowel cancer are good examples. We are even beginning to identify possible genes for schizophrenia, manic depression and dementia. Ultimately of course almost every trait and disease that we have will have some underlying genetic basis, if not as its absolute cause, it's likely that the susceptibility to a particular condition will be due to underlying genetic determinants.

continued on page 2



Dr Anand Saggarr MBBS FRCP
Consultant in Medical Genetics



'The London Clinic has built its reputation on delivering excellent clinical care and for attracting many of London's leading consultants.'

Malcolm Miller, Chief Executive

Noted

One Gene or Two? continued

Welcome

Our consultants are referred the most complex and challenging cases and Mr Colin Davis, Consultant Obstetrician and Gynaecologist demonstrates this when he talked to us about a recent patient who was given a second chance to have children.

Last year we launched a new service – Cardio-Pulmonary Exercise testing. In this edition of 'the Notes', Dr John Goldstone, Consultant Anaesthetist and Intensive Care Lead for The London Clinic presents the results of his recent audit. We also have Mr Sam Singh, Consultant Orthopaedic and Trauma Surgeon answering questions regularly asked by GPs on the diagnosis of sprained ankles.

Mr Marcus Setchell, Consultant Gynaecologist and Obstetrician, Professor Roger Kirby, Consultant Urologist and Dr Peter Amoroso, Consultant Anaesthetist, recently completed a five-day trek across Jordan to raise funds for their respective charities. We would like to offer our congratulations on their achievement and are delighted to include a report on their trip.

We are keen that our newsletter is continually interesting and educational, so if there are any topics you would like to see included in future issues please send us an email to info@thelondonclinic.co.uk with your suggestions.

Malcolm Miller
Chief Executive, The London Clinic



So where do we go to with all of this genetic information? From a patient and family point of view access and delivery of information are extremely important elements of a clinical genetic service particularly given that genetic diagnoses and the potential risk to individuals, the possibility of impact on their life and occupation or the birth of an abnormal child are extremely stressful times and patients often wish to be seen quite urgently. There are many sources of information, not least of all the Internet, but this is not always accurate and often alarming. Families need clear, reliable and compassionate advice.

Clinical Genetics, as a speciality, is an important part of that information giving resource together with making the diagnosis and trying to identify what other problems are associated with the condition and what the future might hold. Furthermore Clinical Geneticists are often an excellent conduit for linking to both patient groups and researchers.

Ultimately medical genetics is about the patient and is trying to take us from the gene to the molecule to the organ and how that fits in with the body in general. Therein lies the continuing hope for the future.

Situations where Genetic diagnosis, investigation and counselling is advisable

Children with a congenital abnormality, inherited condition in their parents, developmental delay or dysmorphic syndromes

Adults affected by congenital abnormality or inherited condition

Parents who have lost a child or a stillborn baby with a congenital abnormality or an inherited condition or who have suffered reproductive loss (termination for abnormality or recurrent miscarriages)

Individuals with a family history of

A known or suspected genetic disorder or

A common condition with a genetic component including familial cancer or

Urgent assessment of a pregnant woman with abnormalities found on prenatal scanning.

Mental retardation

Abnormal physical sexual development
Amenorrhoea

Dr Anand Saggar MBBS FRCP
Consultant in Medical Genetics
Appointments: 0870 7518827



'These first results are extremely encouraging. By testing high-risk patients with CPX we are able to significantly reduce complications and even reduce mortality from essential surgery.'
 Dr John Goldstone



'Uterine fibroids and endometriosis can be treated with laparoscopic surgery ensuring a much speedier recovery for women.'
 Mr Colin Davis

John Goldstone reports on the positive outcomes of his CPX audit

NCEPOD identified the high morbidity and mortality following major surgery. In the UK, approximately 500,000 major procedures are performed, most of whom tolerate surgery well and have few complications. However, when cardiopulmonary reserve is limited and undetected, there is a high mortality which usually occurs within the ITU many days post surgery. Typically, such patients are frequently readmitted following surgery with a combination of sepsis and organ failure. Can we identify the patients with a limited reserve, and what is the effect of treatment? The publication 'Modernising Care for patients undergoing Major Surgery' was published in 2005, to summarise the 'state of the art'.

Cardio-pulmonary exercise testing (CPX) is the best method of screening patients prior to major surgery and enables patients to be triaged into high, intermediate and low risk groups. Such an approach enables the high risk patients to receive pre-emptive treatment on the ICU. This includes fluid therapy and inotropic support, peri-operative beta blockade and other therapies to optimise the patient and this dramatically decreases the cardiovascular risk following surgery.

CPX is now available as an outpatient procedure. It involves mild exercise on an upright bicycle with continuous ECG and metabolic monitoring. We have audited the introduction of CPX at The London Clinic at the end of 2005.

After 50 patients we assessed the results. 16 CPX tested patients were in the high risk group. During the same time interval, 223 patients were audited who had not been screened with CPX.

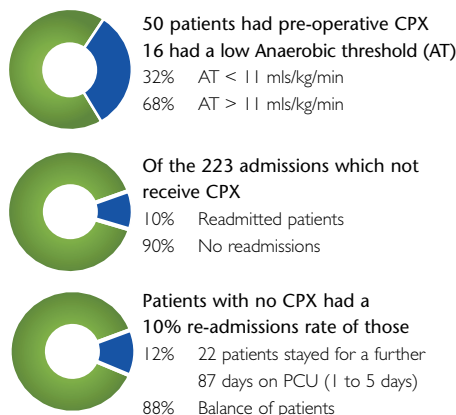
Of the 50 screened patients, no patients were subsequently re-admitted, no patients developed cardiovascular complications and all patients were discharged to the nursing floor. This contrasted with the patients who were not screened, 12% of whom were re-admitted to ICU, almost all of which had cardiovascular complications post surgery.

Of the 22 patients who were re-admitted following high risk surgery, all developed organ failure, and a further 87 days of ITU was required, followed by a prolonged length of stay at ward level care. The mortality was 5%.

CPX is fully supported by the private medical insurers. To arrange a CPX test contact Sally Dadds, the CPX nurse on 07738 004461.

Dr John Goldstone MBBS MD FRCA
 Clinical Director of Intensive Care Medicine
 Tel 07973 182548

CPX: The first 50 tested patients



The London Clinic helps woman keep family dream

A 40 year-old scientist from Kuwait who developed uterine fibroids was given a second chance to have children thanks to a complicated procedure carried out by Mr Colin Davis and his team at The London Clinic.

Uterine fibroids are benign tumours of muscle and connective tissue that develop within, or are attached to, the uterine wall and can result in heavy periods and infertility.

Back in her homeland, the patient was seen by two gynaecologists whose only answer to the problem was to perform a hysterectomy. The patient's strong desire to have children led her to The London Clinic, in search of another expert opinion.

Mr Colin Davis, a consultant obstetrician and gynaecologist recommended a myomectomy rather than a hysterectomy. The operation, although not unusual, is often highly complicated and has up to a 5% risk of the patient requiring a blood transfusion.

The operation involves removing the fibroids from the uterus, after which the uterus is repaired and 'closed'. This requires a high level of expertise and is best performed by a fertility specialist to ensure that the woman's chances of conceiving in the future are enhanced.

Mr Davis comments: "Common gynaecological conditions such as uterine fibroids and endometriosis can be treated with laparoscopic surgery ensuring a much speedier recovery for women. This particular patient was discharged from hospital after five days with her chances of having children being significantly improved".

In conjunction with The London Clinic, Mr Davis is pioneering the introduction of robotic surgery for use in myomectomy procedures in the UK.

Mr Colin Davis MBBS MRCOG MD
 Consultant Obstetrician and Gynaecologist
 Tel 020 7616 7753

Persistent pain after an ankle sprain represents a difficult clinical problem with patients often presenting late to their General Practitioner. This article explores causes of persisting ankle pain and its management.

Ankle injuries managing the persistently painful sprain



Mr Sam Singh MA FRCS (Orth)
Consultant Orthopaedic and Trauma Surgeon
Appointments: 020 7616 7693

What are the ligaments of the ankle?

The ankle has three main ligaments- the lateral ligament, the medial deltoid ligament and the syndesmotic ligaments. (Fig 1) The horizontal anterior talofibular ligament, part of the lateral ligament complex, is most commonly injured and is palpated by starting at the tip of the fibula and moving anterior while applying an inversion or inwards twist to the ankle.

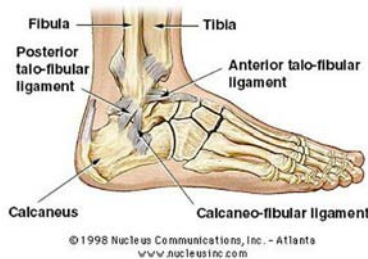


Figure 1

How useful is clinical examination of the acute sprain given most of the ankle seems to hurt?

The ligaments of an acutely swollen ankle are difficult to assess. It is worth checking the medial ligaments as medial tenderness is associated with a fracture. Two other bony landmarks that should be palpated include the base of the 5th metatarsal and the proximal fibula, as both are also fractured by an inversion injury. A high fibula fracture may be the final result of a twisting force that has torn all the ligaments and the interosseus membrane holding the tibia and fibula together.

When should we worry about an ankle sprain?

In an acute injury, excessive pain, deformity and the inability to weight-bear are indications for an x-ray. Most sprains can be adequately managed by RICE- Rest, ice, compression and elevation. Pain and swelling persisting beyond two weeks may be from a more severe soft tissue injury, a missed fracture or joint cartilage damage. These patients may benefit from referral to an Orthopaedic Consultant.

How does physiotherapy help in persistently painful ankle sprains?

Physiotherapy is the first step in restoring patient's confidence and stability in their ankles. Local modalities help control swelling while functional rehabilitation strengthens muscles around the ankle that can replicate some of the function of the ligaments. Balance exercises such as the wobble board teach patients to recruit these muscles when they feel their ankle is giving way.

What is ankle instability and why is it important?

A patient may fall over if their knee is unstable. In the ankle, however it is more subtle. It may just be that they experience pain on exercise or lack confidence when walking in certain shoes or on uneven surfaces, feeling happier indoors than in a park or on the golf course.

Recurrent sprains restrict recreational activities and they are at a high risk of cartilage damage and arthritis in later life. Unlike the hip or knee, most ankle arthritis is a direct result of trauma.



CONSULTANT PROFILE

Mark Ho-Asjoe qualified from the Royal College of Surgeons in Ireland in 1989. After finishing his housemanship, he began his training in surgery and won the first prize gold medal in the Part I examination of the FRCS.

His early basic surgical experience started at St Mary's Hospital, London, followed by specialist training in Plastic Surgery at Mount Vernon Hospital. After further training at St Andrew's Centre for Plastic Surgery and two years spent in basic science research, Mark began his higher surgical training in Plastic Surgery in the Pan London rotation.

In 2001, Mark furthered his training in oncological and microsurgical reconstruction at the world renowned centre, Chang Gung Memorial Hospital in Taiwan, one of the biggest microsurgical reconstructive Plastic Surgery centres in the world. His training in both microsurgery and Asian aesthetic surgery continued at Queen Mary's Hospital, Hong Kong, where he was appointed as Honorary Clinical Associate before returning to the UK. The completion of his Plastic Surgery Specialist training in 2002 was complemented by an aesthetic fellowship at the Wellington Hospital.

In 2003, Mark was appointed Consultant Plastic & Reconstructive Surgeon at St Thomas' Hospital, London with a special interest in microsurgery and breast reconstruction. His other interests are in lower limb trauma and aesthetic surgery.

Mr Mark Ho-Asjoe
MB Bch FRCS FRCS(Plast)
 Consultant Plastic, Reconstructive and Aesthetic Surgeon

Suite 1, 14 Queen Anne Street,
 London W1G 9LG
 Tel 020 7079 0057
 Fax 020 7079 0051
 Email: mhoasjoe27@yahoo.com

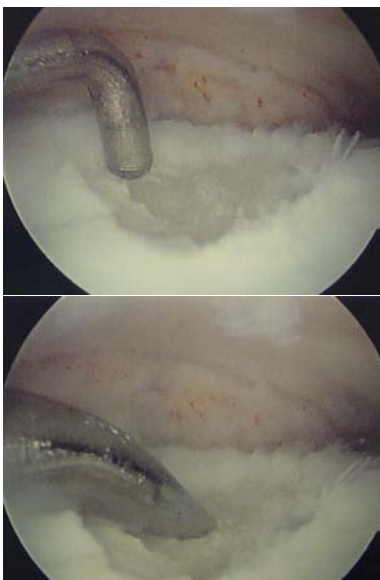


Figure 2



Figure 3



Figure 4

When do I operate for ankle instability and what does surgery entail?

Surgery is considered when after non-operative treatment, usually in the form of physiotherapy or bracing has failed to improve the patient's symptoms. Further investigation with an MRI is useful to exclude other causes of ankle instability or pain.

Significant joint pathology such as cartilage defects or excessive scar tissue can be treated arthroscopically. At surgery the lesions are debrided and the hard subchondral bone is 'micro fractured' with a pick. This causes inflammation and bleeding which stimulates protective junk cartilage formation (Fig 2).

My lateral ligament reconstruction of choice is a Brostrum procedure. In this the remnant of anterior talofibular ligament and calcaneofibular ligament are detached from the bone with a cuff of tissue (Fig 3). They are advanced and reattached in a bony groove (Fig 4). This tightens the ligament and treats the instability.

Figure 2
 The lateral ligament complex of the ankle. The upper arthroscopic image shows a large osteochondral defect of the talus after debridement. This is treated with microfracture (lower image) to stimulate bleeding in the local bone and laying down of scar tissue.

Figure 3
 The first stage of the Brostrum lateral ligament reconstruction involves taking of the ATFL and CFL as a thick cuff off the bone.

Figure 4
 A bone channel is made in the distal fibula. The cuff of tissue which includes the ligament is now advanced, reinforced and reattached. This treats the instability.

Consultants granted admitting /practising privileges

from January 2006
to May 2006

Cardiology

Dr Stuart Harris
BSc (Hons)II MBBS MRCP
Consultant Cardiologist
NHS post(s): King George Hospital and
St Bartholomew's Hospital
0795 829 3310

Dermatology

Dr Ellen Francis Lawlor
MB.BCH.BAO DCH D.OBST RCOG M.R.C.P.I MD
FRCPI FRCP
Consultant Dermatologist
NHS post(s): Newham Hospital
020 7363 8004

Endocrine surgery

Mr Fausto Palazzo
MS FRCS(Gen)
Consultant Endocrine Surgeon
NHS post(s): Hammersmith and Charing Cross
Hospital
020 8383 4004

Gynaecology

Mr Khalil Razvi
MRCOG FRANZ COG
Consultant Gynaecologist
NHS post(s): Southend and Basildon Hospital
020 8445 0370

Mr Paul Carter
Bsc (Anatomy) MBBS FRCS MRCOG FRCOG
Consultant Gynaecologist
NHS post(s): St Georges Hospital, London
020 8672 1255

Haematology

Dr James Cavenagh
MB BS MRCP MRCPATH MD FRCP FRCPath
Consultant Haematologist
NHS post(s): Barts and The London NHS Trust
020 7317 2500

Hepatology

Professor Roger Williams
CBE MD FRCP FRCS FRCPE FRACP FmedSci
FRCPI(Hon) FACP(Hon)
Consultant Hepatologist
NHS post(s): University College London Hospitals
020 7616 7693

Neurosurgery

Mr Dan Plev
MD FANCh (Neurosurgery)
Consultant Neurosurgeon
NHS post(s): King's College Hospital
020 7616 7720

Oncology

Dr John Bridgwater
PhD FRCP
Consultant Medical Oncologist
NHS post(s): UCLH
020 7380 9093

Ophthalmology

Mr Hadi Zambarakji
MB ChB FRCOphth D.M.
Consultant Ophthalmic Surgeon
NHS post(s): Whipps Cross University Hospital
020 8535 6508

Orthopaedics

Mr Jason Bernard

MBChB FRCS(ED) FRCS(ORTH) MD(UNIVGLAS)

Consultant Orthopaedic Surgeon (spinal)

NHS post(s): St George's Hospital

020 8672 1255

Mr Samrendu Singh

MA MB BS MRCS FRCS (ORTH)

Consultant Orthopaedic Surgeon

NHS post(s): Guy's and St Thomas'

020 7188 4474

Mr Robin Pollock

BSc MBBS FRCS FRCS (Orth)

Consultant Orthopaedic Surgeon

NHS post(s): Royal National Orthopaedic Hospital,
Stanmore

020 8909 5677

Dr Sean Molloy

DC Doctor of Chiropractic MB BS MSC

(Orthopaedic Engineering) FRCS (Orth)

Consultant Orthopaedic Surgeon (Spinal)

NHS post(s): Royal National Orthopaedic Hospital,
Stanmore

020 8954 2300

Pain management

Dr Dmytro Leschinskiy

MBChB DEAA FRCA

Consultant Anaesthetist

NHS post(s): Queen Elizabeth Hospital

020 8297 4550

Dr James Smart

MBChB FRCA

Consultant Anaesthetist

NHS post(s): University College Hospital

020 7387 9300

Radiology

Dr John Karani

MBBS BSc MRCS LRCP FRCR

Consultant Interventional Radiologist

NHS post(s): King's Cross Hospital.

020 7346 3321

Rheumatology

Dr Shahid Jawed

MB BS MD FRCP

Consultant Rheumatologist

NHS post(s): Kingston Hospital

020 8546 7711 ext 2089

Urology

Mr Krishna Patil

FRCS FEBU

Consultant Urologist

NHS post(s): Ashford & St Peters NHS Trust

0193 287 2000

Mr Omer Karim

MBBS FRCS FRCSUrol MS

Consultant Urologist

NHS post(s): Wexham Park Hospital

0175 362 1815

Mr Christopher Ogden

MBBS FRCS(Ed) FRCS MS FRCS Urol

Consultant Urologist

NHS post(s): Chelsea & Westminster and St. Mary's

020 7603 6067

Mr Mark Feneley

MB BChir MD FRCS(Eng) FRCS

Consultant Urologist

NHS post(s): UCH

020 7636 8333

Amanda Hallums

*Director of Clinical Services,
The London Clinic*





'This is a very exciting time as we come one step closer to realising our strategic aim of building a dedicated centre at the forefront of cancer treatment and care.'

Malcolm Miller
Chief Executive

Plans for hospital development step up a gear

We are delighted to announce the commencement of the next phase of our development strategy to build a state-of-the-art cancer treatment centre in the heart of London's medical community. Formal legal agreements have now been exchanged with Howard de Walden Estate, the Freeholder, relating to the development of the site on the corner of Marylebone High Street and Devonshire Place.

Demolition of the existing buildings on the site is now under way and the construction of the new purpose built hospital will begin later in 2006. It is anticipated that this phase of development will open in early 2009.

Approaching an investment value of £50M, this multi-million pound facility will rank amongst the world's best and will house state-of-the-art equipment and facilities. It will significantly improve the quality and scope of treatment and services available to patients in the capital.

Some of the UK's most eminent consultants and healthcare staff will work in the centre, which will provide radiotherapy, chemotherapy, Haematology transplant facilities and other specialist oncology services.

This structure will enable us to consolidate our existing comprehensive range of cancer services as well as providing the opportunity to develop new services in a purpose built environment.

The Clinic's aim is to deliver excellence in individual care to our patients in accordance with our charitable objectives and this development is another example of how we are able to reinvest to achieve this aim. The cancer treatment centre will contain 47 inpatient rooms and 24 day care beds/chairs.

Malcolm Miller, Chief Executive of The London Clinic comments; "The London Clinic is committed to provide excellence in healthcare and the new hospital will allow us to continue to lead the field in cancer services. This is a very exciting time as we come one step closer to realising our strategic aim of building a dedicated centre at the forefront of cancer treatment and care. It will house the latest technology and equipment and offer progressive treatments to cancer patients."

Hike for Hope

The London Clinic's very own Mr Marcus Setchell, Professor Roger Kirby and Dr Peter Amoroso recently donned their walking boots for the trek of a lifetime in aid of WellBeing of Women (WoW) and the Prostate Research Campaign UK.

Professor Kirby and his colleague Marcus organised the five-day trek and managed to encourage 100 other fellow fundraisers to join them as they made their way from the Dead Sea to Petra in Jordan, taking in the breath-taking scenery of the Arabian desert along the way.

The group flew to Amman and started the arduous task of trekking from Mount Nebo, through the Jordan Valley, Desert Wadis and rocky mountains until they reached the spectacular 200 year-old red rock city of Petra. Here they finally hung up their boots and celebrated their achievements.

Professor Kirby says: "This was a truly amazing experience. We were so proud of everyone who took part and completed the mammoth

journey. We are sure the magnificent views along the way and reaching the stunning city of Petra more than made up for the gruelling challenge".

The group raised a staggering £700,000 from the event, all of which will go towards raising awareness and improving treatment for both men and women's cancers.

The Prostate Research Campaign UK is the only national charity dealing with all forms of prostate disease, benign and malignant.

WellBeing of Women funds the finest gynaecological and obstetric research that translates into new clinical treatments and brings benefits to women worldwide.



Trustees of the London Clinic
20 Devonshire Place
London
W1G 6BW
www.thelondonclinic.co.uk
Tel +44 (0) 20 7935 4444
Fax +44 (0) 20 7486 3782
Email info@thelondonclinic.co.uk