

Between January and April 2011, CRY screened over 3,500 people with family memorial screenings being the majority of these.

Over 1,000 people were screened at 8 family memorial screenings. These were held in memory of Tom Reid (Garforth Community College), Neil Wickers (Darwen Access Point), John Marshall (Edge Hill University), Craig Lunt (Hilton Hotel, Isle of Man), Martyn Luckett (Parkhouse Centre, Bude), Oliver Hewitt (Aldridge Methodist Church), Allyn McCluskie (Filey School) and Eve Linforth (Summerfield GP & Urgent Health Centre, Birmingham).

Sports screening included ASA Swimming, GB Rugby League, GB Gymnastics, England Rugby Under 20's, Under 18's, Under 17's and Under 16's, Indian Gymkhana Club, Lancashire County Cricket Club, GB Hockey, Warwickshire County Cricket Club, The Royal Ballet Company and GB Netball. Over 650 athletes were screened.

931 young people were screened at 9 school screenings. These were held at Canford School (Wimborne), Bryanston School (Dorset), Nottingham High School, Old Swinford Hospital School (Stourbridge), Tiffin School (Kingston-upon-Thames, held in memory of Neil Desai), Bishop's Stortford School (held in memory of Caroline Johnstone), Millfield School (Somerset), Kings School (Bruton) and Eton College (Windsor).

At our regular CRY clinics, a total of 645 young people were screened. Clinics were held at Colchester General Hospital (in memory of Andrew Gard), the University of Glamorgan (in memory of Mark Stephen Young) and at the University of Ulster Jordanstown and Coleraine campuses. The clinic at the University of Ulster included screening the Ulster Rugby squad.

At the ICAP clinics, where those born in 1995 in the south east are able to get free screening, 247 youngsters were screened. These screenings are held at the CRY Centre for Inherited Cardiac Conditions and Sports Cardiology in St. George's Hospital.

What happens at a screening?

The basic test is an electrocardiogram (ECG) which is a simple non-invasive and painless test that examines the electrical activity within your heart. The ECG involves lying down quietly and it only takes 5-10 minutes. Small stickers are placed at strategic points on your chest, arms and legs. Flexible leads (known as electrodes) that extend from the ECG machine are then attached to these stickers. The electrical rhythm of your heart is recorded and printed out. This part of the process only takes 2-3 minutes to perform.

The ECG printout is then reviewed by a doctor in conjunction with a personal and family history questionnaire.

If a more detailed image is needed (about 5% – 10% of individuals), an echocardiogram (ECHO) can be taken – this is similar to the ultrasound scan that is used for a pregnant woman to check the health of her baby. Soundwaves echo against various parts of the heart and they are recorded on a screen. This provides a detailed picture of your heart's structure and how well it is functioning. This takes about 30 minutes to perform.

CRY uses top of the range machinery donated by Philips for the screening programme. The screening programme is under the aegis of Professor Sanjay Sharma.

Report from the **CRY Centre for Cardiac Pathology** (CRY CCP)

By Jemma Wells, Dr. Mary Sheppard's Medical Secretary at the CRY CCP.

The CRY Centre for Cardiac Pathology received 67 sudden death cases during January to April 2011, an increase from the 47 cases referred during the same period in 2010.



Dr Mary Sheppard
Consultant Cardiac Pathologist



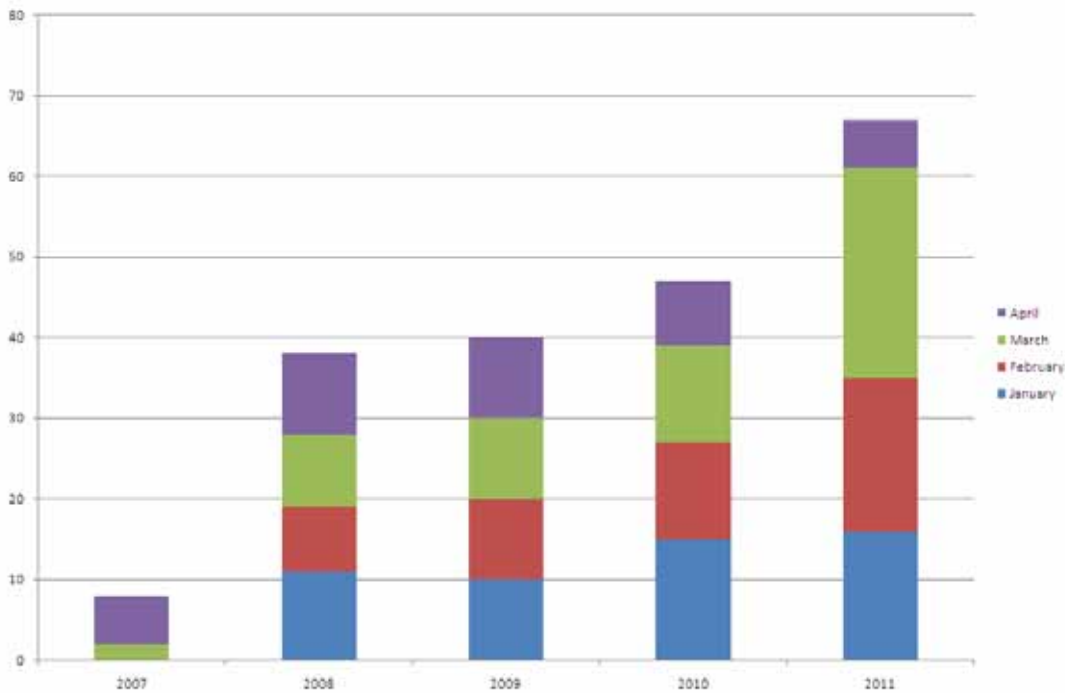
Jemma Wells
Medical Secretary



Dr Sofia de Noronha
Research Assistant



Sahamaz Vakhshouri
Laboratory Technician



Despite the number of referrals rising, we are pleased to report that our average turnaround time is still less than 14 days, with only the exceptional case taking a bit longer. In 2010 we achieved an overall average turnaround of 9 days and we aim to maintain that average in 2011 and continue to provide a fast-track service.

As shown in the pie chart above there are a variety of causes of sudden cardiac deaths, this chart shows the diagnoses that were made during January – April 2011. The majority of our cases are sudden cardiac death with a structurally normal

April saw a small decline in numbers however this was due to the Easter break and bank holidays that fell within the month and this was counterbalanced by the increase in referrals during the first 3 months of the year, with more than double the number of cases received in March compared to the same month in 2010. This is shown in the figure below.

heart indicating most likely electrical abnormalities. Once a diagnosis is made we provide a report to coroners and families.

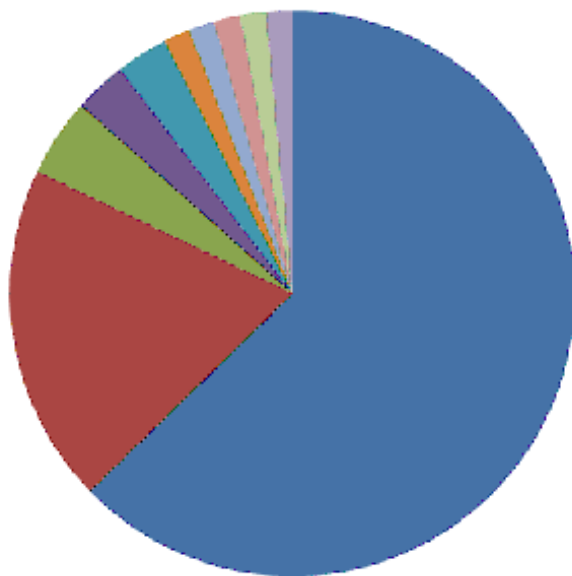
Genetics

In April 2011, CRY Research Fellow Dr Hari Raju got his pilot genetics study underway in collaboration with the Royal Brompton Hospital and St George’s Hospital. A special solution, RNAlater, allows tiny samples of splenic tissue to be preserved at room temperature enabling the tissue to be sent by courier along with the heart – previously the spleen had to be stored in the freezer at -80°C and transported using dry ice. The sample can be used for potential future genetic testing on behalf of the family providing consent is given.

In cases of sudden cardiac death where Dr Sheppard diagnoses a possible inherited cardiac condition, genetic analysis may sometimes be used to confirm the exact cause of death, identify the genetic mutation responsible, and determine which surviving relatives may also be at risk, and hence benefit from treatments to reduce that risk.

Research

Regarding the research in our unit, results of our fast track cardiac pathology service for victims of sudden cardiac death (SCD) in the UK have been presented as oral presentations at international conferences including the United States & Canadian Academy of Pathology (USCAP) in San Antonio, US. This work has also been



- Congenital Heart Disease
- Ischaemic Heart Disease
- Infection of the heart
- Coronary Artery Disease
- Valve Disease
- Sepsis
- Transplant vasculopathy
- Aortic disease

accepted as a poster presentation by the European Congress of Cardiology taking place this summer in Paris, France. The paper reporting these results is being finalised. We are also currently analysing all cases of sudden cardiac death referred to Dr Sheppard in the last 17 years, approximately 2,000 cases, the largest database in the world.

We recruited BSc student Katsuya Norita, a third year biomedical student, to investigate coronary vasculitis, an inflammation of the coronary arteries in the heart which is a rare cause of sudden cardiac death that can affect young people. He has recently submitted an abstract of his findings to the European Congress of Pathology to be held in Helsinki, Finland, this summer and he is now preparing the manuscript.

Specialist registrar Dr Anna Silvanto has recently submitted her research on heart attack with normal coronary arteries in sudden cardiac death (SCD). This work was presented as an oral presentation at the Speciality School Pathology Annual Trainee Conference in the Royal College of Pathologists and accepted as a poster presentation for the British Division of the International Academy of Pathology (BDIAP) and the Pathological Society of Great Britain & Ireland meeting in Ghent, Belgium, in May.

Dr Sofia de Noronha has recently obtained ethical project approval to investigate arrhythmogenic right ventricular cardiomyopathy (ARVC) using genetic and microscopy analysis. This project will involve a close collaboration with the Genetics Research Laboratory within the Cardiovascular Biomedical Research Unit at the Royal Brompton Hospital led by Professor Stuart Cook and Dr Paul Barton.

Other collaborative projects in our unit include a clinico-pathological investigation of channelopathy by CRY Research Fellow Dr Hari Raju and cardiology consultant and senior lecturer at St George's University, Dr Elijah Behr.

Six articles were published in peer-reviewed journals in the first quarter of this year. These included:

1. Carpenter JP, He T, Kirk P, Roughton M, Anderson LJ, de Noronha SV, Sheppard MN, Porter JB, Walker JM, Wood JC, Galanello R, Forni G, Catani G, Matta G, Fucharoen S, Fleming A, House MJ, Black G, Firmin DN, St Pierre TG, Pennell DJ. On T2* Magnetic Resonance and Cardiac Iron. *Circulation*. March 2011
2. Patel J, Sheppard MN. Sudden death owing to right atrial hemangioma. *J Forensic Sci*. 56 (2):529-530 March 2011

3. Chenn SS, Chaparro-Munoz M, Uemura H, Gatzoulis MA, Sheppard MN, Kilner PJ. A multilobulated intracardiac mass in Ebstein's malformation. *Eur Heart J* March 2011
4. Deshmukh M, Francis N, Galinanes M, Ang K, Muller S, Sheppard MN. Primary cardiac low grade sarcomas mimicking myxomas. Pitfall in diagnosis. *Diagnostic Histopathology* April 2011;17(4):206-209

February saw the launch of two important endeavours in inherited cardiovascular disease. Firstly, Dr Sheppard teamed up with the Dean of the Royal Society of Medicine, David Misselbrook, to host an expert forum to discuss genetic testing after sudden cardiac deaths. On the 3rd February key figures including coroners, cardiologists and pathologists and the CEO of CRY, as well as members of the Human Tissue Authority met to discuss the importance of genetic testing and outlined a possible protocol that could be introduced UK wide.

Secondly, Dr Sheppard travelled to Doha in Qatar to attend a research focus meeting on Cardiac Genomics, athlete's heart and inherited cardiovascular disease at the Qatar Orthopaedic and Sports Medicine Hospital. The meeting was attended by an international team of specialists in molecular cardiology, sports cardiology and inherited cardiac pathology. Both of these meetings are looking towards a future where genetics will play a vital role in preventing cases of sudden cardiac death.

However, it is essential that our future pathologists are well trained and well read on the new developments in this field. Dr Sheppard feels strongly about the provision of well-rounded training at a high standard for future pathologists and so she attended the BDIAP seminar for trainees in Histopathology at the Royal College of Pathologists to deliver a lecture on sudden cardiac death.

Two specialist cardiac pathologists retired this year and will not be replaced. This highlights the need for specialist training in cardiac pathology.

