



The Histiocytosis
Research Trust
together we'll find a cure

H R TRUST NEWS

Welcome to the first edition of H R Trust news, the new bi-annual newsletter full of the latest updates about The Trust, latest medical research, the Circle of Friends and all the different fundraising initiatives taking place.

The Trust is dedicated to promoting and funding scientific research into uncovering not only the causes of LCH and HLH but also ensuring early diagnosis, effective treatment and a cure. We also want to support patients and their families as well as raise public and professional awareness of histiocytic disorders.

We especially want to hear from you – your news, your stories and your fundraising ideas – whatever you want to say, please get in touch!

With best wishes for Christmas and the New Year from all of us here,

Shelley
Trust Co-Ordinator

Trust Funds Three Year £327,000 LCH Project



Professor Matthew Collin

THE Histiocytosis Research Trust's latest research project will focus on LCH and improving diagnostic and prognostic tests as well as therapeutic targets. Professor Frederic Geissman of King's College, London and Professor Matthew Collin, of the Institute of Cellular Medicine at Newcastle University will be leading the study.

The duo's project, entitled Cell Autonomous Mechanisms in the Pathogenesis of Langerhan's Cell Histiocytosis, was selected from 12 applications received after the Trust invited applications for the Jon Pritchard Research Award last May.

Professor Collin said: "We are delighted to receive this award. It will allow LCH research in the UK to take a unified stance against the disease with clinicians and scientists all working together. With LCH IV and the help of all the patients, we hope to be able to look at the cells and tissues of LCH in new ways, to explore exciting new ground related to the recent discovery of the V600E BRAF mutation in the majority of LCH cases."

Nikolas Symposium a Landmark for LCH

THIS year's Nikolas Symposium has been hailed a landmark as clear targets were set at which to aim new drug treatments for LCH. The conference was entitled Dendritic Cell Genomics and Metabolomics: Clues for LCH Causes and Cures and was attended by scientists and doctors from all over the world, including H R Trust Trustees Dr Vasanta Nanduri and Professor Peter Beverley.

Professor Barrett Rollins of Harvard University, who had attended a previous Nikolas Symposium, identified in 2010 a change in the DNA in half of LCH samples he examined. This specific change called V600E, has been seen in melanoma, colorectal and thyroid cancers.

Since the discovery of V600E in LCH is a breakthrough in understanding the cause of LCH, it also indicates that LCH is a form of cancer providing a clear target for drug development.

Delegates discussed methods for searching for further mutations and also heard a preliminary report from a pharmaceutical company conducting a clinical trial into a new treatment for LCH.

HLH Project Striding Ahead

Scientists Bobby Gaspar and Adrian Thrasher together with Dr Marlene Carmo are making great strides in the Development of Gene Therapy for Familial HLH which is being funded by the H R Trust at the Institute of Child Health, London.

The team are looking at replacing Perforin, the commonest gene to cause inherited HLH. They have discovered that in order to cure HLH, patients who have an abnormal gene need to be given a normal perforin gene. To achieve this, viruses called lentiviruses are used to carry the normal gene into the patients' cells.

Cells called stem cells are taken from the bone marrow and infected with the viruses. When they are put back into the patient, the stem cells make white blood cells and these should make normal perforin and prevent the patient getting HLH, when they have a virus infection. It is also important that the perforin gene that is given to the cells is properly controlled, so that perforin is only produced in fully developed white blood cells, not in the stem cells. To do this, the researchers are working to insert into their lentiviruses, control elements, that will prevent them producing perforin in the stem cells.

The researchers have made several different lentiviruses to find out which one works best to produce perforin in white blood cells but not stem cells. They tested these first by infecting cell lines lacking perforin and measuring how much perforin was produced. The best virus was then used to infect stem cells from mice that have an abnormal perforin gene, just like HLH patients, and the stem cells were then put back into the mice.

Two months later white blood cells from these mice were shown to produce normal perforin, although in quite low amounts. However, the white blood cells were very good at killing virus-infected cells in tissue culture, suggesting that they may have enough perforin to prevent HLH.

The researchers are now carrying out an exciting experiment with collaborators at Cincinnati Children's Hospital. Perforin deficient mice are given stem cells infected with the lentivirus carrying the normal perforin gene. The mice are then challenged with a virus called lymphocytic choriomeningitis virus. This causes HLH in perforin deficient mice and the object of the experiment is to see if mice given the stem cells carrying the normal gene are protected from HLH. If the experiment works and the mice are protected, it will be an important step forward in the development of gene therapy for perforin deficient HLH patients.

LCH Parent and Patient Road Show

An LCH Adult and Paediatric Parent and Patient Road Show took place recently at the Leeds General Infirmary, Leeds. Speakers included Drs Vasanta Nanduri, Johann Visser, Kevin Windebank, Professor Matt Collin and Professor Peter Beverley. Attendees also had the opportunity to speak to them individually. To find out more, check out www.hrtrust.org

Volunteers Sign Up to Help Trust



Anne Devenney

A GROUP of volunteers has stepped forward to help move the Trust forward and raise funds.

Sherrie and Paul Bennett, Anne Devenney, Adrian and Lynn Oldfield, Samantha and Andrew Rowley, Joanna Savva, Stephen Masser and Hilary Bickler have all been allocated specific roles as part of an initiative to improve communication, support, administration and most importantly fundraising. If you would like to get more involved with The Trust, then contact Shelley at Shelley.ross@hrtrust.org



Stephen Masser

A Busy Year for Artemis

It has been a busy year for the Greek-based Artemis Association, the sister organisation of The H R Trust.

In May, athletes Andriani Fouseki and Nikos Korras travelled from Greece to take part in the BUPA London 10,000 to raise funds for The Trust. Members of Artemis, through a variety of fundraising activities, have also contributed greatly to funding The Trust's latest fundraising project, Cell Autonomous Mechanisms in the Pathogenesis of Langerhan's Cell Histiocytosis.

And just recently runners, including Nikos V Kontoyannis, nephew of the President of both Artemis and The H R Trust, Paul Kontoyannis, have taken part in the 29th Classic Marathon Race, in Greece.

Meet ...Dr Vasanta Nanduri



Dr Nanduri is a paediatrician at Watford General Hospital and is an expert in paediatric and late effects LCH as well as HLH. She is a member of The Histiocyte Executive Board and is co-chairperson of The Histiocyte Society Epidemiology/late effects study group. She is married with two children.

Vasanta trained to be a paediatric doctor in India and has worked at a number of hospitals in the UK including Northwick Park, Hammersmith, Edgware and Barnet as well as University College. In addition to being based at Watford General Hospital, Vasanta runs an LCH paediatric clinic

It was while working at Middlesex Hospital in the paediatric oncology and endocrinology departments that Vasanta first met the late Dr Jon Pritchard and her interest in LCH began. Dr Pritchard was instrumental in setting up The H R Trust with current president Paul Kontoyannis.

“Jon used to say I had a beam in my eye when looking after LCH patients and I became his trainee. I completed two years of clinical oncology, a year and a half of endocrinology and then undertook four years of research into LCH.

“I would hope that we will have a cure for LCH within the next 10 years but the problem is we still do not understand why or how it happens,” she explained.

Vasanta is hoping a BPSU HLH Epidemiology Study will go live in the next six months. The study will look into the epidemiology, incidence, causes and genetic profile of HLH in children in the UK.

The study, which will cost approximately £20,000, is being funded by Jason and Kelly Carter.

Vasanta attended her first Histiocyte Society meeting with Dr Pritchard in 1996 and since then has attended every meeting. She was elected onto the committee in 2000 and has been a member of the Board for the last six years.

When she is not looking after her patients, Vasanta enjoys spending time with her family and cooking.

Stephen is an inspiration



Trust volunteer Stephen Masser started displaying symptoms of LCH when he was a baby, but was not finally diagnosed until the age of two. However, he has never let his symptoms hold him back despite the difficulties he has faced along the way.

While growing up, Stephen remembers being in and out of hospital as he kept breaking out in rashes which would not disappear. Doctors also discovered that his pituitary gland was affected and as a result, he had to undergo regular hormone injections.

At the age of 16, Stephen stopped treatment. However, despite feeling that LCH has blighted both his childhood and even his life as an adult, he is determined to make the most of things.

He explains: "LCH has affected my whole life. It has affected my body, my growth and my make-up. I had learning difficulties at school and made friends with people who were quiet.

"I was always going back and forth to hospital. Mum always had to look after me and I was never left alone. I felt left behind at school and never bonded with anyone."

However, since leaving school, Stephen has been involved in the Territorial Army for seven years and is currently working in a warehouse. However, he hopes to train as a paramedic and has recently been accepted on to the training programme.

The 25-year-old has also been appointed by the Trust as an LCH Befriender and plans to raise funds for the Trust.

Stephen can be contacted at smasser@hotmail.co.uk

Stephanie's Miraculous Journey



STUDENT Stephanie Savva was like any other 19-year-old enjoying her first year at university when she woke up one night in excruciating pain. Doctors diagnosed a urine and kidney infection and she was given a 14 day course of antibiotics.

However, several weeks later, Stephanie's neck began to ache and continued to hurt for several days. She was convinced she had slept badly and bought herself a heat wrap thinking this would rectify the problem. However, whilst visiting her boyfriend, she became increasingly exhausted and felt unwell. Her knees itched, her neck ached and she couldn't stop shaking. Although, her symptoms receded, they returned a couple of days later along with a fever.

Back at home, Stephanie began to feel slightly better, but it was while ice-skating with friends, she realised how unsteady she had become and her legs felt weak. Her temperature shot up and on the way home, friends commented on how pale and yellow she looked. She also developed a rash on her face and felt extremely cold.

During a family trip to Cyprus over the Christmas holiday, Stephanie became more and more unwell. Her parents called out the emergency doctor and she was given Tamiflu tablets as well as an injection to reduce her temperature. However, on the way home, Stephanie's eyesight began to deteriorate and she felt nauseous.

Back home, Stephanie went to hospital for blood tests but within hours was recalled and advised that her kidneys and liver were failing and was diagnosed with possible septicaemia (blood poisoning). Stephanie thought she was going to die.

Stephanie's heart and lungs were also now affected and she was transferred to the Royal Free Hospital where she was eventually diagnosed with HLH driven by the Epstein Barr Virus.

Stephanie was treated with a large dose of chemotherapy and her family were warned that she had a 50 per cent chance of survival. However, after three months, she was discharged from hospital but continued to be treated as an out-patient. She relapsed twice but since October 2010, Stephanie has not undergone any treatment and is making excellent progress. She has returned to university and is trying to lead as normal a life as possible.

She explains: "I'll never forget this experience as I am sure it will continue to be the most important in my life. I know I will never be cured especially if I relapse but it has shown me that life is short and to live with no regrets as we don't know what is round the corner. It has taught me to be grateful for everything I have but also to remember that the only thing that really matters in this world is to be healthy, happy and safe with your family and friends around you."

Scientist's Contribution to Seeking a Rational Cure Immeasurable

H R TRUST Scientific Review Board member, Dr Ralph Steinman, died at the end of the September following a lengthy battle with pancreatic cancer. He was also a member of the Nikolas Symposium Steering Committee and was awarded the 2011 Nobel Prize in Physiology and Medicine on October 3rd.

Canadian-born Professor Steinman was a member of the Scientific Review Board between 2004 and 2011 and played an extremely active role in reviewing the applications made to conduct research into LCH and HLH. His advice and recommendations were immeasurable.

Despite being unwell during the calls made in 2008 and 2010, Dr Steinman continued to provide perceptive criticisms and greatly helped the SRB to determine which application would be awarded funding.

He was also a valued member of the Nikolas Symposium Steering Committee and his guidance and support will be deeply missed.

Dr Steinman was awarded the Nobel Prize in Physiology or Medicine "for his discovery of the dendritic cell and its role in adaptive immunity".

H R Trust President Paul Kontoyannis said: "We express our condolences and sincere gratitude to Ralph's family for his steadfast dedication to our cause and for his many contributions to helping us understand more about Histiocytosis which will enable us to seek a rational cure.

We have lost not only a great and dedicated scientist, but also a wonderful friend."

Become Part of Olympic History

Places are still available in next year's BUPA London 10,000 on Sunday May 27th. By taking part you will enjoy a one-off opportunity to experience the Olympic course before the official Olympic athletes do. If you would like to take part, please contact Shelley at shelley.ross@hrtrust.org or by calling 07850 740241.

Fundraising

The Trust is reliant on fundraising to ensure that we continue to fund vital research projects and support patients and their families. There are a number of fundraising ideas and tips on the website – www.hrtrust.org – and you can always talk to one of our team, should you require further advice and support.

In addition to inviting friends and family to sponsor you on JustGiving, you can now also set up a page on Virgin Money Giving. Virgin Money Giving do not charge The Trust a monthly fee, so more of the money you raise, goes towards funding research and support.

Together We'll Find a Cure

Thank you:

- Jason and Kelly Carter for raising £16,751.05 which will fund the BPSU Surveillance Study in memory of Esme Carter
- Howard Ingleby and Jason Miller for raising £10,145.58 after taking part in the Skyline London to Paris Cycle Ride
- Joe Elliott for raising £5,000 from a curry evening and for taking part in the Bank of America Chicago Marathon
- Speedy Hire for raising £4,004.10 after employees ran in the BUPA Greater Manchester Run
- The Wiltshire Family for raising £4,040 in memory of Tilly
- Brett Hill for raising £3,222.83 after cycling from Workington to Sunderland
- Nicola Wilkes for raising £1,516.26 after taking part in the Great Midlands Fun Run
- Staff from YPO for raising £1,295 after cycling from Morcambe to Filey
- The Scotting Family for raising £1,076 in memory of David
- The MacKay Family for raising £1,120 from their annual Garden Party
- Debbie Moseley for raising £1,029.60 from a sponsored weight loss event
- Steve Nolan for raising £931 after taking part in the Surrey Slog Cross Country Half Marathon
- Sherrie and Paul Bennett for raising £830 after organising a Family Fun Day and Netball Tournament
- David Graham for raising £610 after taking part in the BUPA Great North Run
- Emma Krug for raising £500 after organising a Spring Fair
- Lisa Wolf for raising £500 after competing in the Adidas Women's 5K Challenge
- Owen Thoms for raising £483 after taking part in the BUPA Junior Great North Run
- Anne and John Devenney for raising £400 after taking part in the Great Midlands Fun Run
- Dennis Cutting and family for raising £353.10 from an annual bingo evening in memory of Maureen
- Michael Chater for raising £345 after organising a disco
- Sarah McCormack for raising £336.53
- Rawlett Community Sports College for raising £305.72 from a variety of sponsored activities
- Barry Apperley for raising £265 after taking part in the Cardiff Half Marathon
- Emma Holloway for raising £240
- The Hibbard and Crocker Families for raising £200 in memory of Allison Crocker
- Beth and Mike Betty for raising £200 from an annual cricket tournament
- Simon Harper for raising £197.44 after taking part in the Mazda Blenheim Triathlon
- Tessa Bridgeman for raising £193.54 after taking part in the BUPA Great Manchester Run
- The parents of Yvonne Weeden for raising £174 from their Golden Wedding celebrations
- Nick Randall for raising £150 after showcasing the Sue Randall Memorial Chopper at motorcycle shows
- Steve Darlaston for raising £149.77 after taking part in the Edinburgh Marathon
- Moseley Round Table for making a donation of £100

If you have raised money for the Trust recently and you do not see your name, please get in touch and we will include your contribution in the next newsletter.

Meet some of our Fundraisers



Anne Devenney
"By dressing up as Iggle Piggle, we managed to raise greater awareness about HLH."



Brett, Katie and Josh Hill
"We have had amazing support with our fundraising and I would like to thank everybody."



Emma Holloway
"I would like to raise at least £500 for the Trust which has been very supportive to my future brother and sister in-law. "



Winners of Sherrie Bennett's netball tournament. "The day went extremely well and we had great support from everybody!"



Nicola Wilkes
"I feel very privileged and honoured that I have represented this worthy cause on and hope to make a little difference."



Howard Ingleby and Jason Miller
"We need £380K now to fund some very exciting research projects to help us find a cure."



Students from Rawlett Community Sports College with Anne Devenney who said: "They all did amazingly!"



Joe Elliott
"I am trying to raise funds for this worthy cause, but I am also trying to raise awareness of it and what it can do to people's lives"



Nick Randall with the Sue Randall Memorial Chopper
"I will showcase the Sue Randall Memorial Chopper as a vehicle to raise money for The H R Trust."



Staff from YPO, Wakefield
"Employees at YPO are committed to raising as much money as possible."

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For more information call 07850 740241 or visit www.hrtrust.org or Email: info@hrtrust.org