News from the UK Lowe Syndrome Trust

JANUARY 2006

Lowe Syndrome Trust

"Care today … cure tomorrow"

Since the January 2005 newsletter , the charity has continued raising awareness of Lowe Syndrome and the need for funding for research which will hopefully lead to better treatments and eventually a cure of Lowe Syndrome, an incurable children's disease. We are delighted to have been able to raise funding to support two new research projects in 2005 for cataract research at the ***Institute of Ophthalmology*** *(Moorfields) and Kidney research at* ***Addrenbrooke's Hospital Cambridge****.*

All the families affected by the disease are extremely grateful for your continued support and especially those recently diagnosed. . Lowe syndrome is caused by a missing enzyme, sometimes a new mutation, the cause unknown. The disease affects the brain, eyes, kidneys, bones and muscles leading to a short life expectancy.

Black and White Ball May 2005

The annual Lowe Syndrome Ball took place on the 21st May at the beautiful Mandarin Oriental Hotel in Knightsbridge. The Theme of the evening was a ***Black and White Ball.***

Tony Hadley *who is Patron of the charity continued to support us by performing a selection of Spandau Ballet hits. Tony kindly arranged for his close friend,* ***Martin Fry*** *(formerly ABC) to perform some of his songs including Poison Arrow and The Look of Love.*

Jono Coleman, *Trust Patron, supported the charity once again by attending the Ball – Jono has also appeared on many TV charity programmes donating his winnings to the charity.* ***The Fabbagirls*** *(Susie Webb and Zoe Nicholas), Lowe Trust supporters from the beginning of the charity, lending their time to so many events, sang a host of Abba songs, intervened with their cheeky humour!. Susie joined the Trust as a Patron approximately 18 months ago.*

Jonathan Ross *(Trustee) recorded a video with* ***Four Poofs and a Piano*** *especially for the occasion and can be downloaded from the website.*

David Prever, *of LBC Radio compered the Auction with great hilarity when handed a breast filler which had fallen out of a dress!!! The Trust is delighted that David has agreed to support the charity and help raise awareness by being a Patron. The evening was a great success raising £25,000.*

Devonshire House School, Hampstead

We have been supported by various schools since forming in 2000 and are delighted that a school which Lowe Syndrome sufferer Oscar Thomas first attended, has taken on the Trust as their charity of the year. The picture shows Oscar age 4 in his Devonshire school blazer.

Grant Thornton Charity of the Year

Following various campaigns to raise the profile of the disease, we were delighted that ***the Lowe Syndrome Trust will be one of two charities named as charity of the year by Grant Thornton.*** *They have organised lots of their own fundraising events throughout 2005 and will continue into 2006.*

Richard Desmond (of Express Newspapers) new charity patron

Richard Desmond *congratulated the Trust on its endeavours to raise the profile of the Lowe Syndrome disease and fund medical research. Richard was delighted to accept an invitation to be charity Patron and in doing so help raise the profile of the disease and charity.*

Lowe Syndrome Research Grant award to Institute of Ophthalmology (Moorfields Eye Hospital London)

A research grant of £50,000 was advertised in December 2005 for research into Lowe Syndrome. Several research proposals were received and reviewed by the Lowe Scientific Review Board who also gained international peer reviews.

An outstanding proposal from the Institute of Ophthalmology to conduct the first research project to investigate lowe syndrome cataracts was approved and a grant of £50,000 was awarded to ***Dr Tim Livine.***

Penny Lancaster *who is also a Lowe Trust Patron, agreed to appear in a publicity photograph and invited Lorraine Thomas (Chair) and Dr Tim Levine to her home. The Press Release was featured in the Daily Express Newspaper, OK! Magazine and fronted the University College London website.*

2005 Flora Light Women's Challenge

Over 25 women ran for the charity in this years 5K fun run and were delighted to have a visit from Skateboarder Dave Cornthwaite who is training to beat the world skateboarding record (more later)

Our thanks go to those who sponsored and attended the fun run on behalf of the charity and in doing so raise just over £1,000 – thank you!

2005 New York Marathon

*We were thrilled to have 8 runners in the gruelling 26 mile marathon this year. At the time of writing this newsletter we are still collecting funds *raised but it looks to be in the region of £20,000 – a staggering amount!– our sincere thanks to* ***Simon Pounds, Richard Middleditch, Craig Rinder, Mark Pringle,***

Lucy Robson, Julie Granger, Simon Johnson and Frances Rayif.

The charity also has places for the ***April 9th 2006h Paris Marathon*** *and.Novem,ber* ***2006 New York Marathon.***

Nigel Wray and Sarracens Rugby Team

The Trust is delighted to have the support of ***Emmanuel Amapakabo "manu"*** *(pictured) of the Saracens Rugby Club, who has introduced the charity to* ***Saracen Director, Nigel Wray****. The Trust recently met with Nigel and are very hopeful of working together in the near future.*

Addenbrooke's Hospital, Cambridge Research Grant Award

Jonathan Ross, *Lowe Syndrome Trust Trustee, awarded a grant to Addenbrooke's Hospital Cambridge. Jonathan is pictured with* ***Dr Anthony Norden and Oscar Thomas aged 12 who has Lowe Syndrome***

Jonathan was delighted to hand over a cheque to Dr Norden which will enable research to continue in the underfunded area of genetic disorders such as Lowe Syndrome.

Anthony has worked in the field for about 15 years and made steady progress towards understanding kidney disease in these patients. But for the help of charity LST, work would have almost ground to a halt and it would be impossible to develop the recent advances made. Hopefully understanding the basic process of kidney protein handling will help develop treatments to protect Lowe patients from worsening kidney disease and even from developing it in the first place'.

Lloyds TSB awards grant to the charity

The Trust continually campaigns for funding to support the office and was awarded a grant of £9,100 from the ***Lloyds foundation*** *to enable the charity to design and print Lowe information booklets appropriate to both medical professionals and families*

The Lottery

The Charity has been shortlisted to receive a grant to cover office costs and were visited by a ***Lottery*** *representative in November. The Trust will be advised in February as to whether they have been successful.*

Spitz Charity Gig

Our thanks to Martin Paling and his R&B band Sheerwarter for organising a charity event at Spitz Wine Bar at Spitalfields Market London in ecember. The evening was a fantastic success raising just under £2000 including donations.

Tony Hadley Christmas Concert

Tony Hadley, Patron, *has once again supported the charity by organising a Christmas Concert at the Fairfields Hall on the 29th December. The event was an overwhelming success with over a thousand excited females (and some men) raising much needed funding to support Lowe syndrome research.*

The charity is also grateful to ***Martin Fry (ABC)*** *and* ***Peter Cox (Go West)*** *and all the musicians and the stage crew for their support by participating in this fun party.*

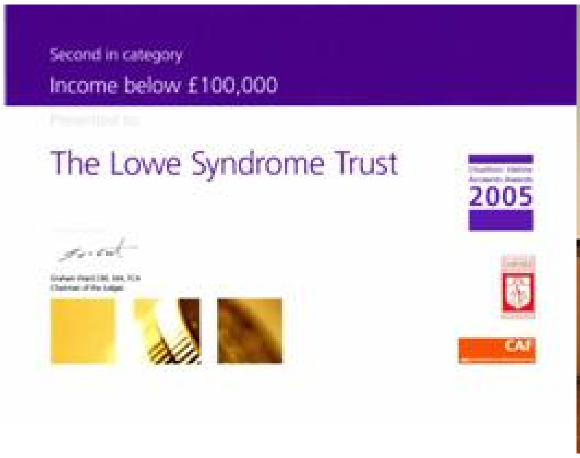
PAYE and Payroll giving

The Trust is continually sending letters to companies re payroll giving. Please see the flyer which we would be grateful if you could post on your company noticeboard!

Lowe Trust Charity recognised by Instutute of Chartered Accountants

The Lowe Syndrome Trust gained another award – this time from the ***Institute of Chartered Accountants and Charities Aid Foundation for openness of accounting .***

A cheque for £750 was presented to Lorraine Thomas and Carolyn Mitchell both trustees of the charity who remarked "we are very excited by this award and congratulations to the volunteers who manage the web site and the accounts with special thanks to Antony Groman. accountants



Second in category
Income below £100,000

The Lowe Syndrome Trust

2005

London to Reims Cycle Ride

The London to Reims Charity cycle ride will take place on 16th June. *We hope to have at least 30 cyclists and a* ***Drinks Reception for all those interested, will take place on 19th January****, sponsored by BT and held at their headquarters near St Pauls. Please contact Lorraine if you wish to attend.*

"Care today … cure tomorrow"
LOWE SYNDROME TRUST

Thank you for expressing an interest in the Lowe Syndrome Trust Charity Cycle Ride 2006.
British Telecom have kindly offered to support the charity by sponsoring an evening when you can listen to a short presentation on the five day event with all your questions answered!

Please join us at 6:30pm on Thursday 19th January 2006
Wine & Canapes will be served!

BT Headquarters
81 Newgate Street
London EC1A 7AJ

RSVP 0208 458 6791 or **lowetrust@homechoice.co.uk** **www.lowetrust.com**

2006 Skateboarding across UK and Australia

Skateboarding Across the World for Lowe Syndrome

In the June 2006 Dave Cornthwaite will attempt the world skateboarding record crossing Australia from Perth to Brisbane.

The previous record was set in 2003 crossing America by Jack Smith, whose son, Jack Jr, died at the age of 14 from Lowe Syndrome. One of the charities Dave will support is Lowe Syndrome.

In May 2006 Dave will warm up for Australia by skating from ***John o'Groats to Land's End (see map)****.*

If any Hotels, Universities, Student Unions, schools, community groups or businesses would like to arrange a fundraising event, or you can offer an overnight stay please Contact Dave through www.boardfree.co.uk.

Other event details to be issued shortly….. The New York Marathon, Cycle Ride from London to Reims, Flora Light Women's Challenge, New York Marathon and other events including a Summer/AutumnBall - we have a top celebrity host lined up for our next Summer/Autumn Ball… full details to be announced shortly or look at our website www.lowetrust.com which is updated continually.

Research

The Lowe Syndrome Trust has organised a 2006 Lowe UK Scientific Meeting when Professor Robert Nussbaum of the National Institute of Health will fly over to meet all the current UK research teams funded by the charity. Also in attendance will be the current Lowe Scientfic Advisory Board team. This will be a fantastic opportunity for an informal meeting to discuss progress to date and a future strategy for Lowe research. The meeting will take place in London on the 17th March.

Lorraine Thomas, chair of the Trust, visited Cambridge in December when she met with Professor Paul Luzio to discuss Lowe Syndrome and research. Lorraine was delighted to be told that the current scientists funded by the Trust are the best within the UK in this particular field.

To highlight all the works of the charity this year would take up too much paperwork, but I would like to take this opportunity of thanking all of our supporters who have made the six UK research grants possible. I would also like to thank the Trustees, Patrons, Lowe Scientific Advisory Board, Andrew for continually working on the website (under much duress!), Judith and Carolyn for their continued dedication to lending one day a week in the office and to everyone who has helped support the Trust

- *Happy New Year!*

LOWE SYNDROME TRUST BBC4 RADIO 4 APPEAL SCRIPT
11 APRIL 2004 PRESENTER: TOM CONTI

Oscar is a cheeky, loveable little boy. A 10-year-old who loves to watch TV, play video games, listen to music – typical of a boy his age. Except that when Oscar was almost six, doctors told his parents that he was unlikely to make it to his seventh birthday. He was diagnosed with Lowe syndrome, an incurable genetic disease that strikes only boys affecting their brain, eyes, kidneys, bones and muscles, leaving the lives of its families drastically changed forever.

One mother whose child died from the complications of the disease summed up her feelings: "I hate Lowe syndrome". I will never forget the moment my husband said those words, on that most horrible of days – the day our beloved son died. I couldn't have said it better myself. What do you do, what do you say, after you have just seen the cold and still body of the child you have loved and tended for twenty years placed in a body bag and carried away."

Struggling to come to terms with the diagnosis and the limited knowledge available, Oscar's mother, Lorraine, decided to set up a UK Lowe Syndrome Research Trust, of which I am proud to be a patron. The good news is that since forming in June 2000, the Trust has raised £200,000 and awarded research grants to Imperial College London, Dundee University, University College London, and Great Ormond Street Children's Hospital.

Another mother, whose son Conor was born with the disease, is so excited that Lorraine has set up the Trust, hoping that the research will help her son live a longer, healthier life. Conor is blind and already has had an eye removed and cannot walk unaided due to the symptoms of the disease including arthritis and swelling of the joints. Sadly Conor is suffering another condition of Lowe which is severe seizures, sometimes having to endure four seizures or more within an hour, which is a most frightening experience,.

Lorraine spends every waking moment campaigning. Doctors feel certain a cure can be found, or better treatment at least, if only their research is supported. It only takes relatively small amounts of funding to continue that research and make a world of difference to Lowe children

THE LOWE SYNDROME TRUST

"Care today….cure tomorrow"

The Lowe Syndrome Trust was founded in June 2000 by the mother of a son diagnosed with the Lowe disease at the age of 5 years. The aim of the Trust is to raise awareness and funding for medical research into the incurable children's disease called Lowe Syndrome. The charity is small and voluntary, organized from a home office but achieving amazing results in terms of funding the first UK medical research into the disease. The NHS has no numbers on Lowe syndrome and there us no Government support and funding into the disease.

The charity has set up a Lowe Scientific Advisory Board and funded six UK research projects to date at GOS children's hospital, Imperial College London, Dundee University, University College London, Institute of Ophthalmology (Moorfields) and most recently Department of Clinical Biochemistry,Addenbrooke's Hospital. In addition, the charity has contributed funding to support three research projects in the USA and organized the first UK Lowe medical symposium December 2004.

What is Lowe Syndrome?

Lowe syndrome was first described by Doctors Lowe, Terrey, and MacLachlan in 1952. It is a rare, progressive, metabolic disease affecting the eyes, brain and kidneys. It may be inherited through the mother as a carrier. Lowe syndrome affects all ethnic groups. **Sadly, life expectancy is short due to the complications and lack of research of the disease**..

What causes Lowe Syndrome?

The cause of Lowe syndrome has been identified as mutation of the OCRL1 gene on the X chromosome. Mutations in the same gene have also been found to cause a different syndrome, Dent disease. Patients with Dent disease have similar kidney problems to patients with Lowe syndrome but do not have the other features found in Lowe syndrome. The exact relationship between Lowe syndrome and Dent disease was a topic at the Symposium and remains to be worked out.

Common features of Lowe Syndrome are:

- *Cataracts in both eyes, found at birth or shortly after leaving the child either blind or partially sighted Severe cases lead to Glaucoma and removal of the eye(s).*
- *Poor muscle tone and delayed motor development*
- *Mental retardation, ranging from borderline to severe with speech, development delay and behavior problems*
- *Seizures (epilepsy)*
- *Kidney involvement ("Leaky" kidneys, or renal tubular acidosis) leading to kidney failure There are cases of kidney transplants as kidneys cause metabolic imbalance resulting in early death*
- *Short stature*
- *Tendency to develop rickets, bone fractures, scoliosis and joint problems*
- *Life expectancy is short due to the complications of the disease.*

What are boys with LS like?
Generally, they are affectionate and sociable, love music, and have a great sense of humour.

How is LS treated?
Treatment of many of the features of Lowe syndrome is symptomatic and may include surgical intervention for cataracts. Speech therapists and nurses specialising in feeding problems can often help. Medication may be needed if children have seizures. Although the use of human growth hormone has been used successfully, careful consideration is needed to balance its use against drawbacks. The renal tubular dysfunction, which causes loss of phosphate, acidosis, short stature, and renal rickets, may be treated by phosphate and bicarbonate replacement therapy.

What about research?
The gene that causes LS was found. In 1995 researchers discovered that the gene defect causes an enzyme deficiency. Supported by the charity, researchers are continuing to investigate the function of the gene and the complicated biochemistry and cellular mechanisms of LS. Other areas that researchers have investigated in recent years include behavior problems and clinical care. **There is no UK government support or research for the disease. The Lowe Syndrome Trust is voluntary and the only UK organization funding research from events and campaigns.**

Can LS be tested?
Reduction of a specific enzyme is known to be involved in Lowe syndrome so diagnosis is made by demonstrating the loss of function of this enzyme and can be carried out by a laboratory test. At present, carrier status can be determined clinically by family history and/or, in most cases, upon careful examination of the lenses of the eye for minor, but nonetheless characteristic, abnormalities. The enzyme test in skin samples is not useful for determining the carrier state. In some families, direct testing of the DNA may also be useful for prenatal testing. In families in which a case of LS has occurred, Prenatal testing should be discussed with the geneticist prior to pregnancy.

Where can I go for more information?
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