



News from the UK Lowe Syndrome Trust
January 2005



Lowe Syndrome Trust
“Care today ... cure tomorrow”



above are friends Charlie and Oscar from North London who both have Lowe syndrome.

Since the July 2004 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 this incurable syndrome. A new Lowe Trust £50,000 grant for medical research has just been advertised which will make it our fifth UK grant - thanks to you - our supporters!. Pictured



*September was the Trust’s fourth year participating in the **Flora Light Women’s Challenge in Hyde Park**. 35 Ladies supported the cause by running/walking the 5K distance and in doing so raising just over £4000.00 for the cause.*



***Penny Lancaster** (girlfriend of Rod Stewart) has joined the **Trust Patron** helping raise awareness of the charity and its endeavours to fund medical research. Oscar who has Lowe Syndrome was delighted to be invited to meet Penny when she performed in the Ben Elton/Rod Stewart Musical “Tonight’s the Night” in London. Penny kindly arranged for Rod to send an auction item for the next LST Ball, a red shirt worn by Rod at a Los Angeles concert – signed by Rod .*

Lorraine Thomas (Chair) with Oscar Thomas and new Trust Patron Penny Lancaster.

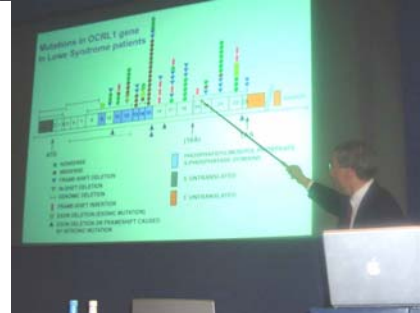
2004 International Lowe Syndrome Medical Symposium at the Royal Society London

Funded and organised by the charity, the first international conference on Lowe Syndrome was held on 2nd December 2004 at The Royal Society in London, followed by a dinner at the RAC club. This historical event brought together over 50 international researchers, medical professionals and a parent to present and discuss the disease, current research underway and ideas for future projects.



*There was a discussion on Dents disease and Lowe syndrome and that the clinical symptoms are not simply accounted for by a DNA test for a single or multiple gene defects as these are the same for both Lowe and Dents. The fibroblast test for a low level of the enzyme (phosphatidylinositol 4,5 biphosphate 5-phosphatase) is a reliable diagnosis of Lowe. **Full details of each presentation can be found on the website www.lowetrust.com***

The meeting was chaired by Prof. Robert Unwin UCL and was opened by Lord Turnberg, Scientific Advisor to the AMRC, followed by Prof. Nussbaum head of the cell biology division of the National Human Genome Research Institute USA. Robert discovered the Lowe's syndrome gene on the X-chromosome and has been instrumental in Lowe Syndrome research. Presenters included Dr. William van't Hoff, Great Ormond Street Hospital, Marianna Mirabelli from Imperial College London, UK and Spain (pictured), Dr John Lucocq from Dundee University, Dr. Martin Lowe from Manchester University, Prof. Chris Oliver from Birmingham University, Prof. Steve Scheinman, Upstate Medical University, USA and Dr. Pedro Cutillas from the Ludwig Institute. Nationalities at the meeting included France, UK, USA, Spain, Germany and Italy.



PROGRAMME

10:00 Welcome Lorraine Thomas, Chair, The Lowe Syndrome Trust

10:15 Introduction Lord Turnberg, Scientific Advisor to the AMRC

10:45 Lowe Syndrome an overview - Prof. Robert Nussbaum, National Institutes of Health, USA (picture >)



12:05 Renal tubular dysfunction in Lowe Syndrome - Dr. William van't Hoff, Great Ormond Street Hospital, UK

12:25 Synthetic receptors for inositol phosphates: a novel diagnostic tool for the oculocerebrorenal syndrome of Lowe - Marianna Mirabelli, Imperial College London, UK

12:45 OCRL1 and protein trafficking at the Golgi apparatus and endosomes - Dr. Martin Lowe, Manchester University, UK (pictured >)



2:15 Lowe Syndrome perspective from a parent - Fiona Fisher from Scotland (pictured right).

2:30 OCRL1 and its lipid products - high-resolution localisation and their role in membrane traffic and cell polarity - Dr John Lucocq, Dundee University, UK

2:50 Behavioural phenotype of Lowe syndrome - Prof. Chris Oliver, Birmingham University, UK

3:30 OCRL1 mutation and the limited renal phenotype of Dent's disease - a 'link' between Dent's disease and Lowe syndrome? - Prof. Steve Scheinman, Upstate Medical University New York.

3:50 Potential insights from urinary proteomics in Dent's disease and Lowe syndrome - Dr. Pedro Cutillas, Ludwig Institute, University College London, UK



4:05 Summing up and open discussion (led by Profs. Nussbaum, Kleta (USA) and Unwin (UCL London) pictured right)



LOWE TRUST PATRON TONY HADLEY WITH THE UK CUP STARS CHARITY MATCH IN ITALY RAISES £8000 FOR LOWE SYNDROME

The Lowe Syndrome charity is delighted to have been supported by the UK Cup Stars Charity football match in Italy. Celebrity team player Tony Hadley a Patron and long standing supporter of the charity presented a cheque to the Lowe Syndrome Trust for £7600.



THE CHARITY MATCH The UK CUP Stars

played in May 2004 in Florence, Italy where they were invited by the Nazionale Italiana Cantanti to play their Italian team in the Artemio Franchi Stadium. The Italiana Nazionale Cantanti are an established organisation that play one big live televised event a year in Italy called “The Partita del Cuore”, which translates as “The match of the Heart”. The Stadium was filled with 45.000 people and another 8 million people watched the match live on TV via Rai Uno. After 90 thrilling minutes the score was 6-6 and penalties were taken. The final score was an incredible 10-10 with the UK CUP Stars winning on away goals!.

The UK CUP Stars team in Italy consisted of the following stars from the music industry: Rod Stewart, Steve Harris (Iron Maiden), Neutrino (So Solid Crew), Tony Hadley and Steve Norman (Spandau Ballet), Jason Orange (Take That), Paul Young, Mick Hucknall, Rick Wakeman (Yes), Chris Evans (radio DJ) and Ritchie Neville, Sean Conlon and Scott Robinson (Five). The football professionals were Paul Gascoigne (Newcastle Utd, England), Ian Rush (Liverpool FC, Wales) and Paolo DiCanio (Lazio Roma, Italy) who played their magic skills on the field for the UK CUP Stars. Team managers were Claudio Ranieri (then Chelsea FC manager) and Joe Jordan (Manchester Utd, Scotland). The Italiana Nazionale Cantanti team played among others: Gianni Morandi, Marco Morandi, Zucchero and Eros Ramazzotti. The ‘Match of the Heart’ raised € 300.000 for charity, of which £8000 was donated to the UK Lowe Syndrome Trust.



Charity Patron Jono Coleman has supported the charity in several TV shows. Jono was on Celebrity Ready Steady Cook for the charity and raised £100 for Lowe Medical Research – Thank you Jono ! Jono will also take part in another Charity programme on Living TV (Feb 2005) for the Charity..... (Jono pictured with Harriet from Heart FM)

The Black and White Ball was January 22nd. The new date will hopefully be made by the end of January. As always, be featured in OK! Magazine.



unfortunately postponed from 21st May. Announcement will be sponsors required – the event will

Lorraine Thomas chair of the Trust continued the campaign of **lobbying government** about the desperate need for help with rare diseases such as Lowe Syndrome. Following meetings with Sir John Pattison, Director of Research and Development for the NHS, Peter Greenaway, Assistant Director of R&D and Lord Warner, Parliamentary under secretary of State, the next appointment was made **with Margaret Hodge Minister for Children**. Although Margaret Hodge confirmed this was out of her remit, she promised to write to the Health Minister for children.



Lorraine also met **with Tim Loughton Conservative Shadow Health Minister and Andrew Mennear, a councillor for Frognal & Fitzjohns Ward** who together were most supportive of the aims of the trust and promised to raise questions about Lowe Syndrome at Parliamentary Question Time (pictured Tim, Lorraine and Andrew).



baroness
susan
greenfield
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The Trust is delighted to report that **Baroness Susan Greenfield** has taken on the role of Lowe Syndrome Trust charity Patron. Susan is a distinguished scientist, broadcaster, write and author of the 'The Human Brain: A Guided Tour' and 'Brain Story' among others. She is Professor of Pharmacology at Oxford, which she holds jointly with her position as Director of the Royal Institution of Great Britain. She has presented numerous TV and radio programmes and is probably best known as "Brain Story" a major six part series on the brain and mind,

broadcast on BBC2 in July 2000.

The Trust will concentrate on various activities this year including campaigns and events such as:-

- The Flora Light Women's Challenge – 5K Race in Hyde Park on Sunday 4th September
- The NewYork Marathon on 6th November
- A Film premier with dinner late Autumn
- A campaign to target companies re charity payroll giving.
- Campaign to work with USA and Europe re research
- Working with Genetic Interest Group
- Lobbying Government
- Trust letter writing re sponsorship/donations
- UK Tax deduction for extra costs of special needs children as in other countries.



**Trustee
Jonathan Ross**

Charity Patrons:

Susie Webb, Tris Payne, Penny Lancaster, Tom Conti, Jono Coleman, Tony Hadley, Richard Park



Some of the children you are helping by supporting this charity ...



THE LOWE SYNDROME TRUST APPEAL

Lowe Syndrome (OCRL) is a devastating genetic disease that affects thousands of little boys born worldwide born with cataracts (either blind or partially sighted), stunted growth, poor muscle tone, rickets, arthritis (some never walk), kidney problems, seizures, epilepsy and mental impairment. In spite of these handicaps the little boys have extremely happy and cheeky personalities. Sadly, few survive to become adults.

Yet today, it may be within our grasp to help cure this disease. The Lowe Syndrome Trust was formed in June 2000 to raise money for medical research to cure this disease. The Trust is small and voluntary organised from a home office, but already has funded Lowe research projects at Dundee University, Imperial College London, University College London and Great Ormond Street children's Hospital – all through campaigns and various fund raising events. All we have to do is raise relatively small amounts of funding.

The disease has no government support or funding for research.



If you would like more information, to attend local events or help sponsor fundraising events, please see www.lowetrust.com or contact the Lowe Syndrome Trust on 0208 458 6791

CALL FOR RESEARCH PROPOSAL IN LOWE SYNDROME up to £50,000

You are invited to apply for research funding from the Lowe Syndrome Trust.

The genetic basis for Lowe Syndrome is caused by a defective gene that results in the deficiency of an enzyme OCRL1 Phosphatidylinositol 4,5-bisphosphate, 5 phosphatase. Lowe's oculocerebrorenal syndrome is a disorder affecting the brain, eyes, kidneys and bones. Research funds are available for funding a research studentship in understanding how the enzyme deficiency leads to malfunction in these specific organs.

Closing date 31st January 2005.

For further information and Application Form please contact Lorraine Thomas, Lowe Syndrome Trust
Telephone 020 8458 6791

(lowetrust@homechoice.co.uk) see www.lowetrust.com

A short note from Anne Brennan, a mother of a son with Lowe Syndrome – Conor

“Hello everybody my name is Anne Brennan. I am the mother of a beautiful little boy called Conor who is 15 years old. Conor has Lowe syndrome. I want to thank everyone who supports the Lowe Syndrome Trust in any way. I also want to thank Lorraine and Andrew Thomas for all the fantastic work they have done with the charity.

Lorraine does an amazing job fundraising and raising awareness for this awful genetic disease. It makes me so happy as a parent, that all these projects are going on to find ways to treat and hopefully one day cure Lowe Syndrome.

I also have two little girls who will one day be tested to see if they are carriers of the disease. I am not looking forward to that day.

Conor is now 15 and has been through so much through the years. How do I tell you about Conor in few words.? Well this is what is happening over the next few days.... today, I took Conor to an orthopedic consultant and I have found out that Conor has a curved spine, a problem with his hip and will need surgery in the next year or two to release pain in these areas and will never be able to walk again. Tomorrow, I bring Conor to an eye doctor to see if his left eye needs to be removed yet (his right eye was removed three years ago) due to infection and the same thing is happening to his left eye. Conor has had 10 operations on his eye (cataracts, glaucoma, corneal graft). He is totally blind in his remaining eye.

Next week Conor will have his blood taken to monitor his renal function. Franconi Syndrome (Lowe)- we always wait with a heavy heart for these results and hope things are not too bad. Renal failure can happen but Conor is taking medication to help treat his kidney condition at this stage.

Conor also suffers with epilepsy, gran mal and partial. He recently spent one month in status (ie his brain was in constant seizure activity). Now he takes four medications to help the condition. He suffers with severe constipation and has weekly enemas. As you can imagine, life can be a struggle... years of looking after this lovely boy have definitely left its mark on us all. The constant worry and anxiety has left us sad and upset lots of time.

But Conor has a very cheeky personality –he always come through the door saying “home sweet home” and is full of fun and mischief – we would not change him for the world.

So I hope you all know how important the work with Lowe Syndrome is and how it keeps us families going”. Thank you all from all Lowe Syndrome families”