

[My son with Lowe syndrome Thomas is a walking miracle](#)

Feeling Lowe

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Hampstead mum, Lorraine Thomas, set up a charity to help find a cure for her son and other little boys with Lowe Syndrome, an incurable genetic disease. Cathy Levy reports



Lorraine and her husband were told that Oscar wouldn't make it to his seventh birthday. He was almost six years old and, their only son, had just been diagnosed with Lowe Syndrome, an incurable disease that strikes only boys and results in short life expectancy.

Thankfully, today, he's almost 10 and when we meet, his birthday is all he can talk about. Sat close up to the television screen in the spacious ground floor kitchen, he's engrossed in a cartoon, only disturbed when Lorraine calls to him to come and say hello. Like a whirlwind, he races over, flings his arms tight around my neck and whispers loudly in my ear: "It's my birthday soon! Have you got a present for me?" He's a cheeky, loveable little boy, like most Lowe Syndrome boys, and again, typically, is very small for his age; closer in size to that of a six year old.

For the past three years, Lorraine has fiercely devoted her life to raising money for the charity she set up to fund research into a cure for the disease. It was first recognised as a disease in 1952 by Charles Lowe, now known to be caused by a defective gene (OCRL1) that may occur with no family history. Boys are born with cataracts in both eyes, hypotonia (poor muscle tone), they suffer from autistic development and speech delay, epilepsy and most damaging of all, kidney problems. "The reason that they die is really from the metabolic imbalance in their body due to the lack of an essential enzyme," says Lorraine, matter of fact. Due to this malfunction, the kidneys dispose of essential amino acids and proteins needed for the body to function effectively, and results in soft bones, rickets, arthritis and poor growth.

After the shock of the diagnosis, Lorraine searched the Internet and discovered there was some hope: "I found out that some of the boys have lived until they're 30," she says. "But most do die in their mid-teens." In the end, the body gives up. Although realistic, Lorraine says she can't think about the future. She repeats these words several times over and I change the subject, to the incredible achievements she's accomplished since launching the charity.

It's taken her on a journey, through a medical world she knew little of before. She began by writing to Tony Blair who passed her on to Lord Hunt who passed her on to Sir John Patterson, director of research and development. After meeting with him twice, although supportive, he told her the government couldn't help, and put her in touch with other charities, except none of these supported medical research. She then set up a scientific advisory board and placed an advert in *Nature* magazine calling for research proposals. These swiftly followed and after a peer review, resulted in several world-leading research projects that have since just begun at Great Ormond Street, Imperial College, University College London and Dundee University.

But these projects could never have got off the ground without the successful fundraising events so passionately organised by Lorraine. They've been featured in *OK!* and *Hello!* Magazines and she even managed to persuade Jonathan Ross to become a trustee, describing him as: "absolutely fantastic and a tower of strength." Patrons also include *NW*'s Jono Coleman, Vanessa Feltz and Tony Hadley.

All the time Lorraine's been fundraising (and it's a full time job, with part time voluntary help from a few committed friends), she's endeavoured to keep Oscar's profile low. She doesn't want him to know about all the bad things that are happening to him. "But time is running out I feel," she says. Doctors have told Lorraine that if they carry on like this, raising enough funds, it really won't take long to find a cure. No wonder then she devotes her life to the charity. The next big event, a spectacular affair, is the Nutcracker Ball being held on 25 November at Six Hamilton Place, Park Lane: a gorgeous venue, with champagne and a three course dinner with wine at just £79/head.

Each year, £50,000 minimum is needed to keep all the projects and research going. Funds also go to the American Lowe Syndrome Association (a support network rather than a fundraising arm, it desperately needs money). For Oscar himself, Lorraine does everything she can including taking him to many alternative therapists. "I work thinking that I've got to get a cure," she says. It's a huge responsibility, but with such drive, determination and driven love, perhaps it's not such an impossible dream after all.

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www.lowetrust.com