The Lowe Syndrome Trust

Care Today… Cure Tomorrow

The Lowe Syndrome Charity

Andrew Thomas – CEO




The Lowe Syndrome Trust

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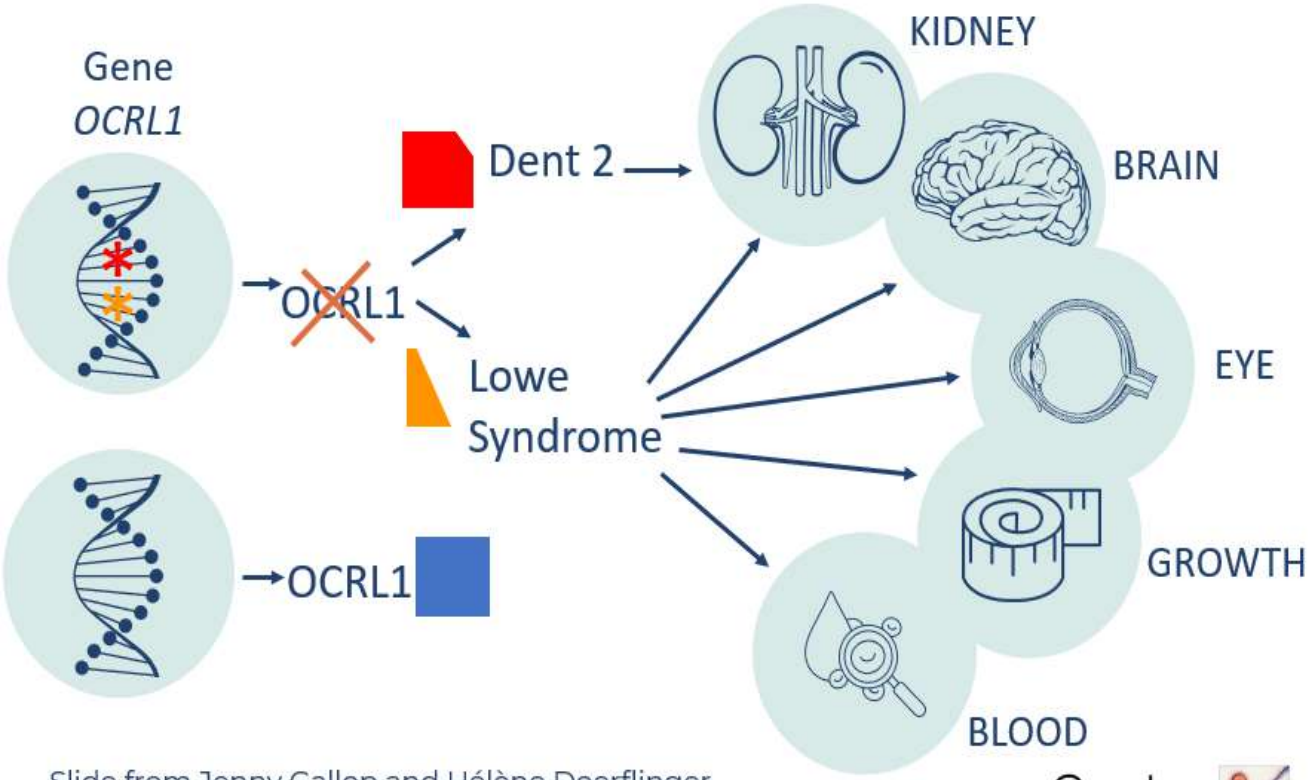
What is Lowe Syndrome?

- Lowe Syndrome is a condition from birth that affects the functions of the eyes, kidneys, brain and general development.
- Eye conditions includes cataracts in both eyes that must be removed shortly after birth. Special glasses and contact lenses may be prescribed.
- Fanconi Kidney condition may result in loss of vital substances and may affect bone and muscle development and may lead to kidney stones.
- Brain condition may include mild learning difficulties and autistic spectrum behaviour.
- Lowe Syndrome is due to a mutation in the OCRL1 gene
    - OCRL1 mutation can also result in Dent-2 that does not exhibit the eye or brain disorders.



Slide from Jenny Gallop and Hélène Doerflinger
University of Cambridge

Gurdon INSTITUTE

The Lowe Syndrome Trust

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- *Founded in 2000 by my late wife Lorraine after our son Oscar was born and diagnosed with Lowe Syndrome, to initiate and coordinate medical research and support families.*
- *Lorraine founded the Trust as a registered UK charity 1081241 with trustees, medical research board, and patrons for fundraising and awareness.*
- *Run by unpaid volunteers, funded by public fundraising and events, grant applications and (currently) a Big Lottery community fund award.*
- *Annual Call for Grant Proposals, typically <£50,000 over 2-3 years, subject to Advisory Medical Board Peer Review Process.*
- *Medical Research meetings held include at the Royal Society London and most recently at TIGEM Naples Italy in May 2023. Perdue USA planned for Oct 2024*
- *UK Family Outreach meetings, 1st held in Cambridge 2023, Durham TBC*
- *Works with organisations including Gurdon Institute Cambridge, LSA USA, AISLO Europe.*

COMMUNITY FUND

The Lowe Syndrome Trust

Care Today… Cure Tomorrow

Core team

Andrew Thomas (CEO)
Anna Bogdanova
- charity coordinator
Anastasia Lanzara
Deborah Harter

Paul James -
Special Advocate

Paul James… Living with Lowes

Medical Board

Prof. Robert Unwin - Chair
Dr. Detlef Brockenhauer
Prof. Shamshad Cockcroft
Dr. Philip Beales
Dr. Peter Cullen
Dr Mike Harrison
Prof. Peng Tee Khaw OBE
Prof. Helen Cross
Dr. Nimalan Maruthainer
Dr. Richard Sandford
Dr. Rudiger Woscholski
Dr Martin Lowe
Dr Jenny Gallop

Trustees

Mr Joseph Laycock - Chair
Mike Fennings
Anastasia Lanzara

Patrons

Jonathan Ross - OBE
Tony Hadley – MBE
Penny Lancaster
Graeme Swann
Lisa Voice
Melanie C
Melanie Blatt
Melanie Sykes
Mathiue Flamini
Mark Emms
Oliver Evans

The Lowe Syndrome Trust

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Collaboration & Knowledge sharing
Symposiums
Medical Meetings

Family Guidance & Support
LSA Facebook
UK Family Outreach
NHS and Social services
Advocacy

News & Publications
- Newsletter
- Summary Leaflet
- Story so far
- Medical Handbook
- Guide to UK Lowe Adult Care (WIP)

Lowe Syndrome Trust
Office & Website
www.lowetrust.com

Governance and Finance
UK Charity Commission
Annual report
Lottery Report
Major Donor Reports

Fundraising events
Campaigns
Marathons
Cycling Tours
Charity Balls

Grant Awards
Medical Board
Grant Review Panel

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Main Research Grants awarded

Year	Institution	Principal Investigator	Research Title
2021	Perdue University USA	Dr Ruben Aguilar	Reactivation of Ocrl1 function in Lowe Syndrome
2020	University of Naples TIGEM Italy	Dr Antonella De Matteis	Testing FDA approved drugs in preclinical models of Lowe Syndrome
2010	University College London	Prof. S. Cockcroft	Membrane targeting and activation of the Lowe syndrome protein OCRL1
2010	Yale University USA	Pietro De Camilli, M.D	Cellular proteins that interact with OCRL1
2008	Imperial College London	Dr R Vilar-Compte and Dr R Woscholski	Detecting PIP2 levels in biopsy samples.
2005	Dundee University	Dr John Lucocq	OCRL1 and its Lipid products – membrane traffic and cell polarity
2005	Addenbrookes Hospital	Dr Antony Norden	Kidney disease OCRL1 and Dent-2
2005	Institute of Opthalmology Moorfelds	Dr Tim Levine	Cell Biological Analysis OCRL1 in human lens epithelial cells
2004	University of California USA	Prof Robert Nussbaum	Mouse Model of Lowe Syndrome
2003	Manchester University	Dr Martin Lowe	Lowe Syndrome Zebrafish Model
2003	Birmingham University	Prof. Chris Oliver	Lowe Syndrome Behaviour
2002	Insitute of Child Health Great Ormond st	Dr Van't Hoff, Dr Robert Unwin, Dr Guido Laube	Proximal tubular dysfunction in Lowe's syndrome

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The Future

- *Plan for charity funding, UK family support and global research*
  - *Support for Lowe and Dents 2*
- *UK family outreach and Adult Care*
  - *Supporting UK Down's Syndrome Act towards better adult and residential care*
  - *Website updated and partnerships with UK NHS, Benefits and Social Services*
- *Closer cooperation with Gurdon Institute, LSA USA, Perdue and other stakeholders*
  - *UK Family Outreach meetings - Cambridge, Durham, Bristol TBA*
  - *Medical Research information and meetings*
  - *Global Patient Registry and Database*
- *Supporting Potential Drug Therapy and Clinical Trials*
  - *Alpelisib – Gurdon Cambridge UK, University of Zurich, Bambino Gesu Hospital, Italy.*
  - *Piclidenoson. – Canfite and TIGEM Italy*
  - *Rapamycin (& statin Rosuvastatin) Purdue University USA*
  - *Pilot clinical trial under ERKNet, the European Reference Network for Rare Kidney Diseases.*

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Thanking you for your support