The patient perspective

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a brighter future for people living with alport syndrome

Susie Gear

UK Kidney Week, Glasgow, Friday 2 May 2014

The patient perspective

1. Facts and figures
2. The patient ‘journey’
3. Alport UK
4. The 2014 International workshop on Alport Syndrome, Oxford
5. How you can get involved

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Alport Syndrome

…is an inherited condition that can cause **kidney failure**, **deafness** and **eye abnormalities**.

Those that inherit it will probably require a kidney transplant when they are **young adults**. It can impact **a large number of people in a family**. It is the second most common form of inherited kidney disease.

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How many have Alport Syndrome in the UK?

450+ on dialysis/transplanted

? How many not on RRT

Carriers – 1000?

Total – maybe 2000?

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Neil Turner Dec 2012

The NHS ‘journey’ for a family with Alport Syndrome

How many NHS hospital appointments a year?

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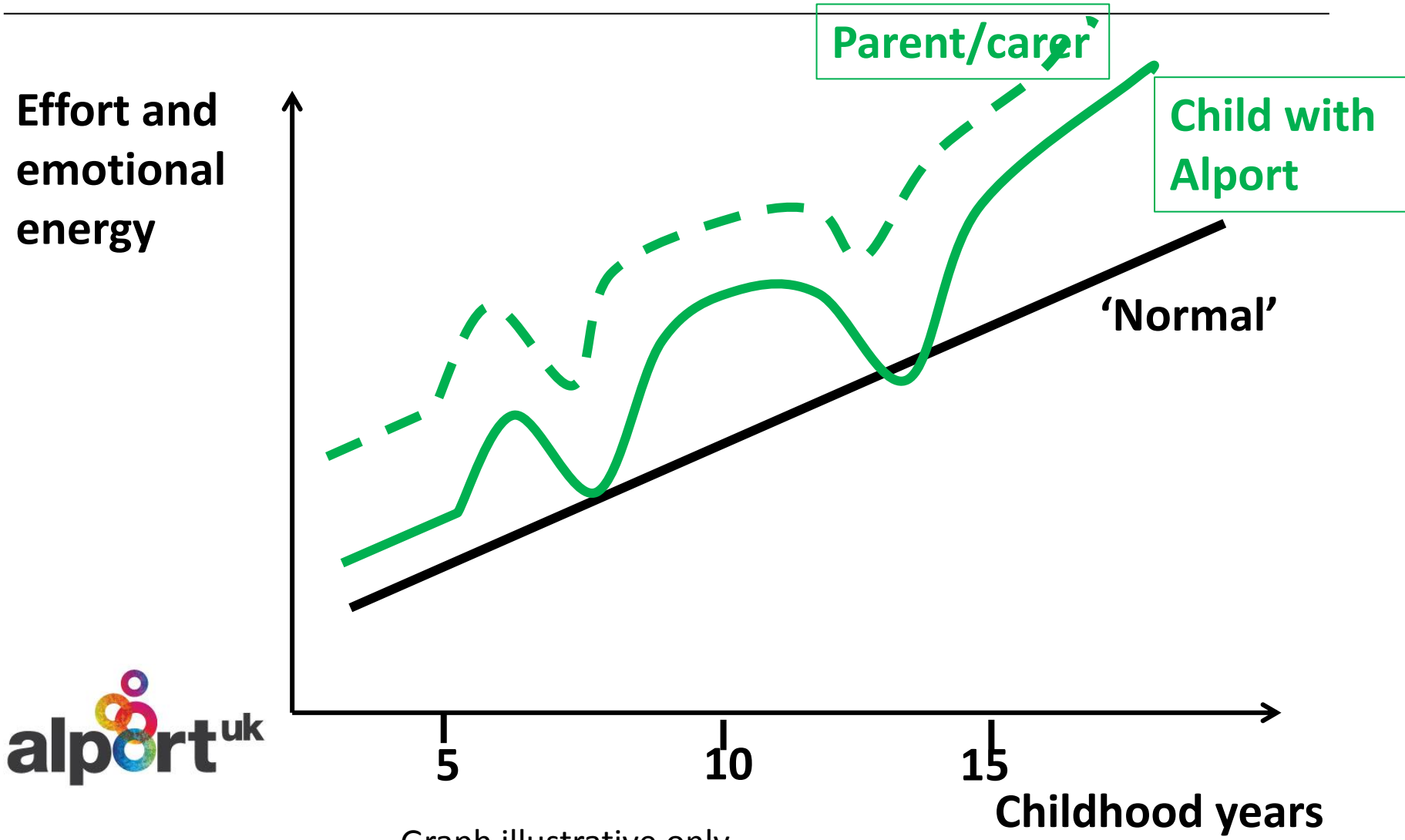
A year in the life of…

- 56 appointments
- 12 different specialists
- £360-400 petrol
- £168 hospital parking
- 224 hours out of school
- 32 days out of working week

In 2014, with one child turned 16, the appointments increased as they aren't necessarily seen together as a family unit.

Alport family with two boys of 15 and 9 years old, 2013

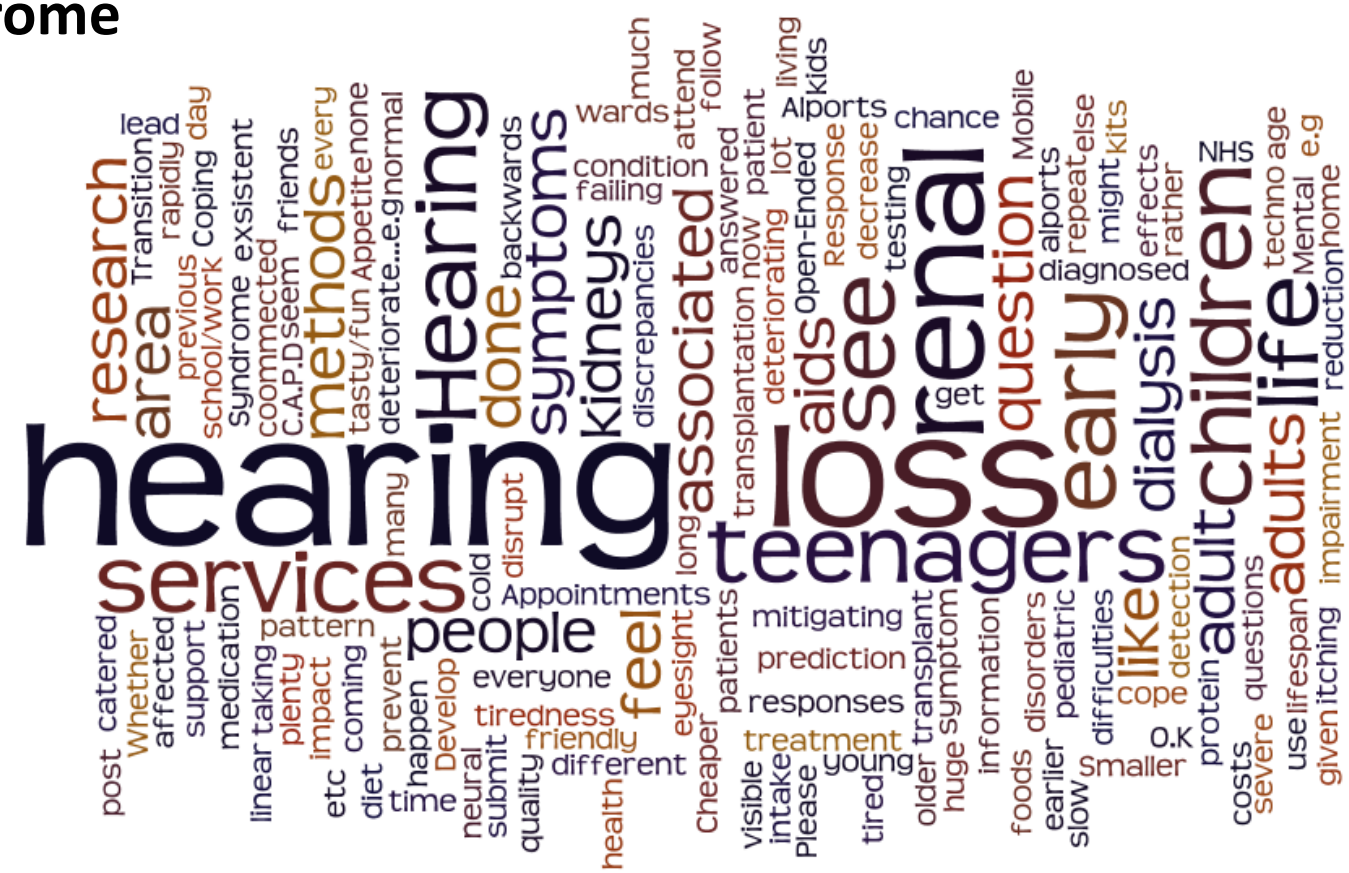
Patient journey – 'normal' vs child with Alport Syndrome



Graph illustrative only

Hearing can have a massive impact on outcome

Q3 Symptoms which affect the quality of life of people with Alport Syndrome



2013 Survey of Alport patients

Patient journeys

- ‘Normal’
- Mother and uncle
- Cousin
- Children
- Carer/parent

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Outcomes and achievements

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We started with an information day in London

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a brighter future for people living with alport syndrome

Charity set up by individuals and families living with Alport Syndrome: Colin Baigent, Katie Fry, Steve Fry, Susie Gear, Catherine Kent, Julie Kiddie, Thomas Kiddie, Clare Norrie, Jules Skelding, Oliver Skelding, Richard Stanton, Julie Tippett, George Walker

In partnership with the Rare Diseases (UK) Working Group for Alport Syndrome.

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Research

Information

Support

Collaboration

A collaboration drawing on different strengths

National **International**

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Other Rare Disease patient groups

2014 International workshop on Alport Syndrome

Workshop organising committee

Professor Colin Baigent, MRC Scientist, Hon Consultant in Public Health, Oxford, UK

Professor Jie Ding, Department of Paediatrics, Beijing, China

Professor Frances Flinter, Consultant in Clinical Genetics, Guy's Hospital, London, UK

Dr Daniel Gale, UCL Centre for Nephrology, London, UK

Professor Dr. Oliver Gross, Department of Nephrology and Rheumatology, Georg-August-Universität Göttingen, Germany

Professor Clifford Kashtan, Professor, Department of Pediatrics, University of Minnesota, USA

Associate Professor Julian Midgley, Department of Paediatrics, Alberta Children's Hospital, Canada

Professor Jeffrey Miner, Washington University in St. Louis, USA

Mr Moin Mohamed, Consultant Ophthalmic Surgeon, St Thomas' Hospital, London, UK

Professor Judith Savige, University of Melbourne, Australia

Dr Judy Taylor, Consultant Paediatric Nephrologist, Evelina Children's Hospital, London, UK

Professor Neil Turner, Professor of Nephrology, University of Edinburgh and Consultant Nephrologist, Royal Infirmary of Edinburgh, UK

Representatives from the following national Alport family support organisations:

Dave Blatt, Alport Syndrome Foundation, Australia

Susie Gear and **Jules Skelding**, Alport support group, UK

Sharon Lagas, Alport Syndrome Foundation, USA

Daniel Renault, AIRG France/Europe

A ‘lock in’ at Said Business School, Oxford on the first weekend in January

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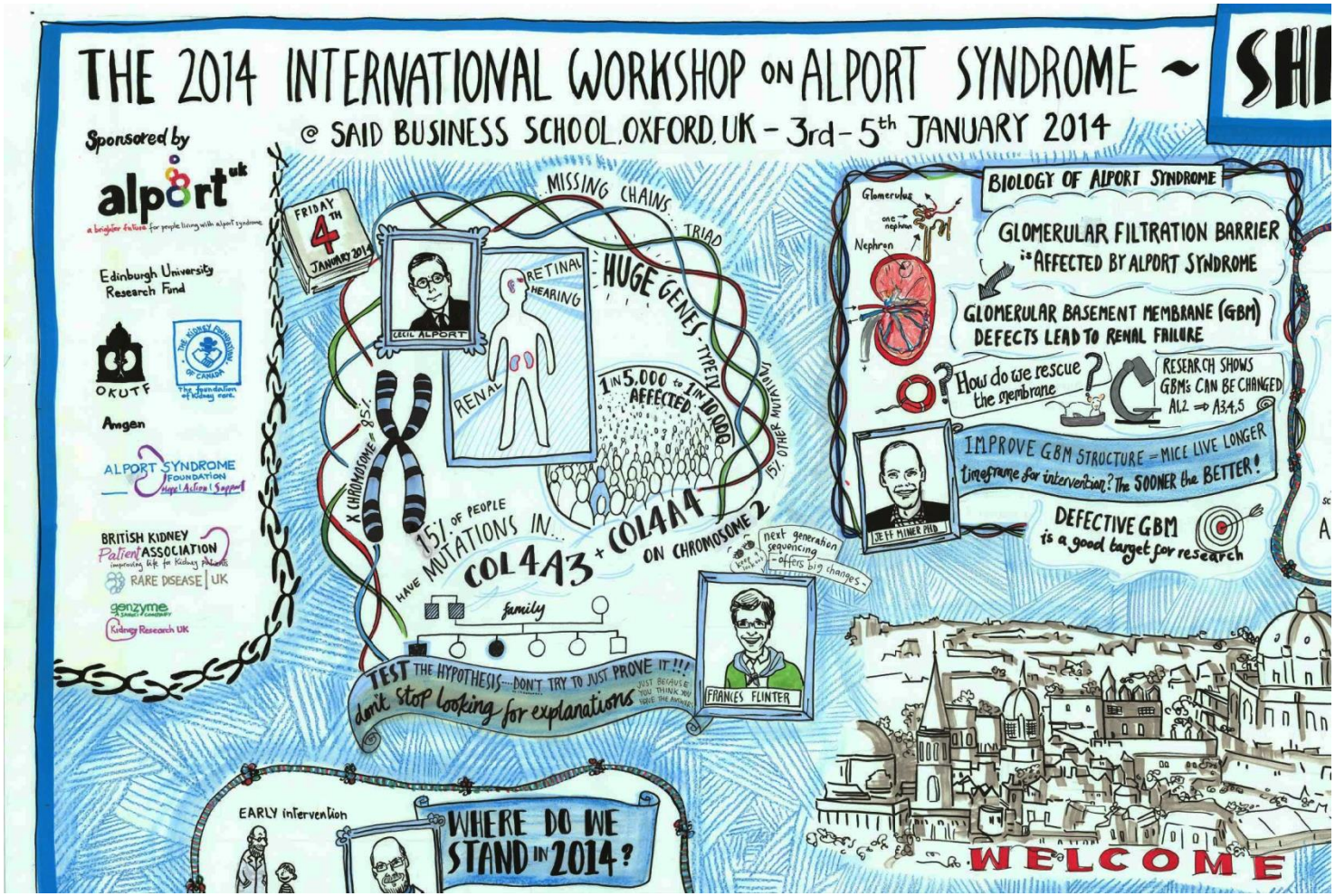
75 people from 17 countries; collaboration even extended to the funding

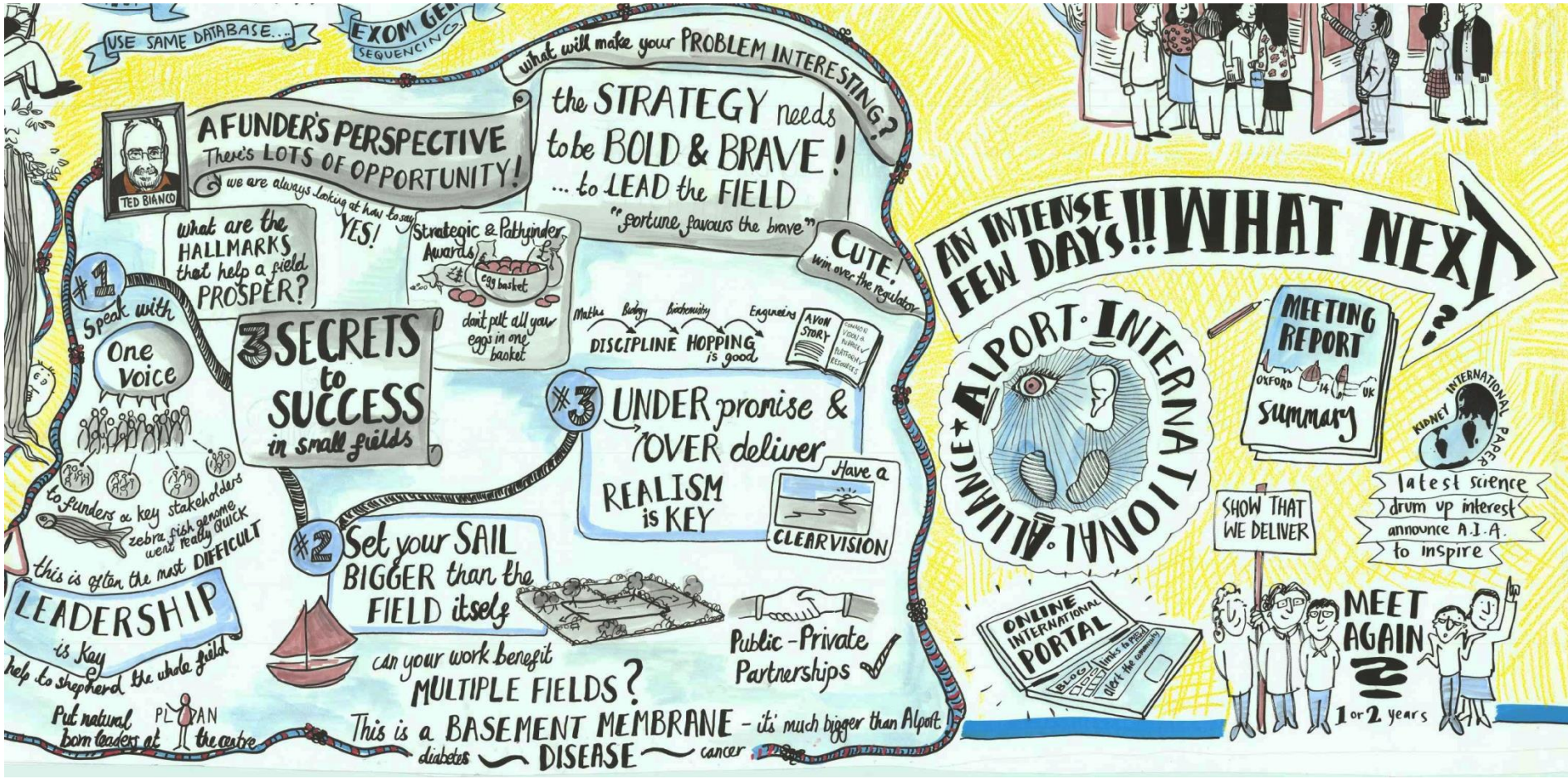


Part of a poster illustrating day 1 of the workshop, created by Endless possibilities

Patients actively engaged in all discussions, including icebergs, evolution and podocytes!

Part of a poster illustrating day 2 of the workshop, created by Endless possibilities

Ted Bianco of the Wellcome Trust inspired us at the end with advice on applying for grants



Part of a poster illustrating day 3 of the workshop, created by Endless possibilities

Outcomes from the 2014 International workshop on Alport Syndrome

- Meeting summary in Kidney International
- Increased number of grant applications for research into Alport Syndrome
  - Kidney Research UK
  - Alport Syndrome Foundation
- Increased collaboration across functional and geographical areas
- Specialist working groups
- Shared need for an International research agenda

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One group that is particularly important for the future

Video

Looking forward to working with you!

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Contact us: info@alportuk.org
Website: alportuk.org