



A charity to promote awareness, research and prevention of adrenoleukodystrophy

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My Family

- Father diagnosed with Addison's disease
- Identical twin sons diagnosed age 6
- Female cousin is a carrier, daughter status unknown
- Male cousin has AMN and two obligate carrier daughters
- Aunt has carrier symptoms

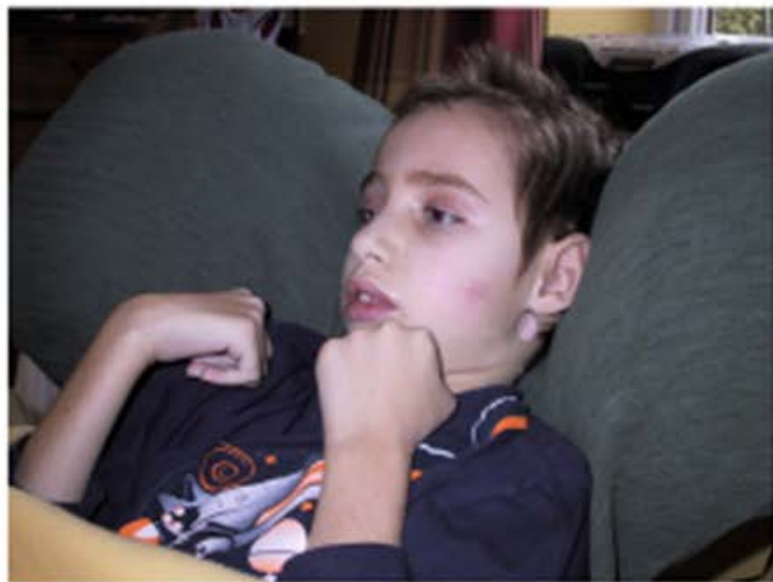
Cameron and Alexander



DISNEY TRIP- 6 MONTHS PRIOR TO DIAGNOSIS



ALEXANDER 6 MONTHS AFTER DIAGNOSIS



Cameron today

- Blind, deaf, no speech,
- wheelchair bound, tube fed
- But still has the best laugh!
- He was 21 in March 2017



Glenn

- Born 2002- in the four weeks between Alexander an Cameron diagnosis...
- Thankfully does not have ALD





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NEWBORN SCREENING UPDATE

NATIONAL SCREENING COMMITTEE

The UK National Screening Committee (UK NSC) advises ministers and the NHS in the 4 UK countries about all aspects of population screening and supports implementation of screening programmes.



*UK National Screening
Committee*

CONDITIONS ALREADY SCREENED

In England, NBS screening is offered and recommended for nine conditions:

- sickle cell disease (SCD)
- cystic fibrosis (CF) (not valid for babies aged 8 weeks or more)
- congenital hypothyroidism (CHT)
- phenylketonuria (PKU)
- medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- maple syrup urine disease (MSUD)
- isovaleric acidaemia (IVA)
- glutaric aciduria type 1 (GA1)
- homocystinuria (pyridoxine unresponsive) (HCU)

HOW AND WHEN

- All screening is optional
- Blood spots are collected on day 5
- Babies can be tested up to a year old if they miss screening
- Screening for Cystic Fibrosis is only offered up to 8 weeks

Cost-effectiveness of including X-ALD in the NHS newborn screening programme Study carried out by



A BRIEF INTRODUCTION TO COST-EFFECTIVENESS ANALYSIS

- Cost-effectiveness analysis is used to help decide whether a new intervention or treatment should be funded
- Decisions in the UK are based on the incremental cost-effectiveness ratio (ICER)
- To calculate the ICER we need estimates of the total costs and health outcomes of X-ALD with and without newborn screening
- The ICER is calculated by dividing the incremental costs of the screening by the incremental health benefits of screening

- Cost-effectiveness analyses are often carried out using decision-analytic models
- These models are able to calculate the incremental costs and QALYs needed to calculate the ICER
- A model enables evidence from a variety of sources to be used together – can include evidence on the intervention, the natural history of the disease, and information about the costs of the intervention and any potential adverse events
- A model also allows uncertainty analyses to be carried out
- A model is a simplified version of reality – cannot capture everything

MODEL PARAMETERS

- Natural History
- Incidence
- Phenotype distribution
- Survival by phenotype
- Survival improvement through earlier transplantation
- Screening parameters
- Sensitivity and specificity of the screening test
- Other conditions identified
- Costs – NHS/PSS perspective
- Costs of screening test
- Costs of long term outcomes
- Quality of life

THE GOOD AND THE BAD NEWS...

- The study showed cost benefit to the NHS
- Application submitted to National Screening Committee April 2016
- Rejected by NSC June 2017 - but with agreement to review in 2019



NSC COMMENTS/CONCERNS

- Uncertainty about incidence and phenotype distributions worldwide and no UK based studies
- Uncertainty about the benefits and dis-benefits for the 78-88% of newborns and their families identified through NBS who will not need HSCT.
- The detrimental effect on families who have a false positive test result
- Identification of other peroxisomal disorders with no current treatment options

NEXT STEPS...

- Collaboration with other groups seeking NBS
- Can we help to change the way NSC views evidence?
- Ensure we meet parameters for any new evidence provided with submission
- Demonstrating “more good than harm”
- Qualitative and quantitative data



GRATEFUL THANKS

- bluebird bio - for their support
- All those involved with NBS in USA - for sharing information
- Well done and congratulations for all of your efforts so far





ALDLIFE

helping to cope - helping to hope

