



# Alström Syndrome UK

## Support Group

### UK MULTI-DISCIPLINARY CLINICS

A Parents journey for improved care for their children.

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Voted: EURORDIS Patient Organisation 2013

# THE DARK AGES

- My son is born 9<sup>th</sup> February 1978
- By 3 months- clear he had problems
- Eyes wobbled (Nystagmus) and he could not tolerate the light (Photophobia)
- Put on weight rapidly
- Eventually referred to an Ophthalmologist
- Diagnosed with Ocular Albinism
- GP states “have another child then you won’t worry about him so much”.
- My daughter is born 11 April 1981
- At 7weeks her eyes wobble and she cannot tolerate the light.



## THE EVEN DARKER AGES

- At 11 weeks my daughter collapses in heart failure.
- Told she will not survive beyond two.
- At 6 she needs bilateral hearing aids
- Both children re-diagnosed with cone-rod dystrophy and Ushers Syndrome.
- Paediatrician says there is no connection between heart and eye problems.
- Multiple hospital appointments are made usually at different times and on different days for each child.
- Parents become professional hospital attendees
- Children have to take lots of time out of school.



# THE BEGINNINGS OF QUESTIONS

- Parents have to fight for everything their children need.
- Decide to study law because I needed to learn how to fight more effectively.
- My son collapses in heart failure with the same condition his sister had at 11 weeks- dilated cardiomyopathy.
- Doctors start to question the diagnosis and look for a condition which connects eyes and heart problems
- Lebers Amaurosis is diagnosed.



# THE PATH TO ENLIGHTENMENT

- Although wrong the diagnosis led to a recall of patients being conducted by a doctor at Great Ormond St. Hospital.
- Alström Syndrome is diagnosed- my son is 18 and my daughter 15.
- We start a charity to find other patients, Alström Syndrome UK in 1998.
- 7 patients attend the first conference.
- The following year we invite the doctors who work with our children so they can see more children.
- Ad hoc voluntary multi-disciplinary clinics begin at the hotel where we hold the conference.



# RECOGNITION AND FUNDING

- The voluntary clinics are moved to a local hospital over a weekend when nobody is using the facilities.
- The clinics continue to grow so we move the clinics to the Centre of England to Birmingham so they are accessible to more of our families.
- After 7 years of organising and running the clinics on a voluntary basis we hear about NHS Funding for Highly Specialised Services.
- In 2006 the hospitals we work with receive funding for both children and adults clinics.
- In 2007 after a long hard fight AS UK receives funding as an equal partner in the clinical



# INTO THE LIGHT

- Funding provides for a professional multi-disciplinary clinic 4 times a year for children and 4 times a year for adults.
- Patients see all professionals on the same day and the professionals communicate with each other for the first time.
- The clinics expand to include a day of tests followed by doctors reviewing the results with the patients the next day.
- Key to the success is the patient-led focus of the clinics-through round table discussions following the clinic.



# PATIENT VOICE

- Patients begin to doubt the experience of the adult clinicians.
- As patients get older increasingly difficult for the current hospital to cope.
- Patients voice is vital to provide evidence to NHS Commissioners of the need to move to a hospital with more facilities.
- Heart, kidney, liver transplants need to be identified much sooner
- Patients and parents become anxious
- The move to a larger hospital is agreed.





# CENTRES OF EXCELLENCE

- Hospitals may not be the ones to advise on this because of the risk of losing their funding.
- Patients voice is paramount
- Competition- the hospital that you have moved from may want to hold on to their expertise and start to compete.
- Need to be constantly reviewed that they are meeting patients needs.
- Patient groups role in ensuring that the Centres of Excellence are truly excellent is crucial.
- Hospitals receive a lot of money for dedicated services- patient groups can monitor that this is spent on the service being funded.



# OUTCOME

- Patients now seen at the brand new state of art hospital- Queen Elizabeth Hospital, Birmingham for adults.
- Birmingham Children's Hospital for our children.
- Patients now tested for all known manifestations of Alström Syndrome regularly.
- Round table discussions with patients at the end of each clinic dictate how future clinics will run and enables the service to remain user led.
- Dedicated Asian mentoring service developed.
- “Excellence is never an accident. It is always the result of high intention, sincere effort, and intelligent execution; it represents the wise choice of many alternatives - choice, not chance, determines your destiny.” *Amelia Earhart*



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