



**Kay Parkinson**  
**CEO and Founder, Alström Syndrome UK**  
**A Parents Perspective**

# Matthew

- Born 9 February 1975
- Normal birth
- By 3 months knew “something was wrong”
- Nystagmus present
- Clinic assured that he was OK and babies eyes were often wobbly in the first few months.
- Very hungry baby and difficult to fill him up

# Matthew

- Concerns increased Matthew taken to GP
- Referred to ophthalmologist
- He had severe eye problems and may have ocular albinism. May have contracted a virus whilst pregnant.
- At 18 months Matthew was diagnosed with congenital dislocation of his right hip.
- One session arranged with a genetic counsellor
- When Matthew was 3 taken to Moorfield Eye Hospital - diagnosed with cone rod dystrophy

# Matthew



# Charlotte

- Born 11 April 1981
- Normal Birth
- Nystagmus seen at 7weeks
- Collapsed in heart failure 11weeks- thought to be caused by a virus. Given a life expectancy of two years.
- Hearing loss found at 6 years
- Paediatrician says no connection between symptoms
- Heart specialist says no need to test Matthew's heart

# Matthew and Charlotte

- For next ten years relatively few symptoms appear.
- Weight is difficult to maintain
- Charlotte is extremely thirsty
- Diagnosis changes as often as specialists, ocular albinism, retinitis pigmentosa, ushers syndrome,
- Both do well at school and Matthew passes the 11+.  
The only grammar school for blind children is RNIB New College Worcester.

# Matthew and Charlotte



# Matthew and Charlotte

- Matthew collapses in heart failure age 16. Given 24 hours to live.
- 18 months later -Lebers Amaurosis diagnosed-referred to Great Ormond Street -
- Miss Isabelle Russell Eggitt knows what has caused the catalogue of problems and diagnoses Alström Syndrome. Matthew is 18 and Charlotte 15.
- Advised to get them both checked for Diabetes which is a known feature of AS.
- They are both found to be diabetic- probably have been for a number of years
- The difference a “correct” diagnosis makes.....

# Alström Syndrome

- We find that there is an International Society of Alström families and research is underway at the Jackson Laboratory in Maine , USA.
- Talk to another mum and hear a mirror image of Matthew and Charlotte's experiences
- Try and find other UK families
- Realise that nobody knows anything about the syndrome.
- Decide to start Alström Syndrome UK charity

# Alström Syndrome UK

- The first Alström Syndrome UK family conference is held in 1998.
- 7 Families attend.
- 1999 Doctors hold impromptu clinics in hotel rooms
- Doctors gain understanding through seeing more patients.
- Patients and doctors work together to develop better care .
- Today we have 50 members. In 2006 National Specialised Commissioning Group fund the clinics and the charity as an equal partner in the service. These are the first clinics for this condition in the world.

# Matthew

- Heart problems increase from 21 years
- Hearing loss diagnosed
- Fitted with 3 different pacemakers
- Problems do not subside and he is referred to Papworth for a heart transplant.
- Transplant doctors would have liked to have seen him 12 months earlier.
- First offer of a heart is received 4 hours after going on the transplant list- later found not suitable.
- Two days after going on the list he receives a heart.
- Soon after the heart transplant he went into kidney failure and sadly died 8 days after transplantation.
- Matthew was 25 years old and a student at the University of Central Lancashire studying social science.

# Matthew (9-2-1978 to 1-5-2003)



# Charlotte

- At 19 operation for an ileal conduit.
- Aged 27 kidney failure and needs dialysis.
- After 5 episodes of pulmonary oedema, dialysis is increased to 4 x per week.
- Charlotte's heart cannot cope very well with the dialysis and starts to fail.
- Referred for a combined heart and kidney transplant.
- Obstacles to listing mean there is a year delay before she goes on the transplant list.
- On 28<sup>th</sup> April 2010 Charlotte gets the call that a heart and kidney are available for her.
- Charlotte died on the 29<sup>th</sup> April 2010 she never awoke from the surgery.

Charlotte ( 11.04.1981 to  
29.04.2010) and fiancé Andre



# Perceptions

- Not what we expected children to be like!
- Not what we expected hospitals to be like!
- Multiple hospital visits-No co-ordination of care
- No information about the disease even when diagnosed
- No medical expertise-No care pathway -No knowledge of what to test
- A&E diagnosis and care-Poor care when in-patients
- Transplant referrals made far too late
- An NHS with no protocols for treating rare diseases
- No adequate social care packages
- Attitude issues- Oh she's now thought of something else that's wrong!

# Matthew and Charlotte

- Ahead of their time
- Trail Blazers – multi-disciplinary clinics set up on the back of the experience of what they went through.
- National Commissioning Group Funding for AS clinics in recognition of the highly specialised services required by patients.
- Family Support now available and medical expertise beginning to grow.
- Big Lottery Medical and Scientific Funding obtained to begin fundamental research into what is happening and why
- BBC Children in Need Funding for children's activities
- Raised awareness of life with a rare disease and helped to instigate change.
- Not what we would have asked for- but a privilege to know.

# Tribunals, Appeals and Courts

- A DSS Mobility Allowance tribunal-successful
- An Education appeal tribunal-successful
- A Disability Discrimination case-successful
- A Medical negligence claim-settled out of court.
- Social service review instigated investigation of treatment of a vulnerable adult whilst hospital in-patient.
- Failure to treat is never cost effective.
- All added burdens on a family already living with high pressures.