



# **The Patient's Perspective and the Role of the Cystinosis Foundation UK**

**Ami Froehlich, Secretary, Cystinosis Foundation UK**

# Early Life



- Born 10<sup>th</sup> October 1985, in Leyland, Lancashire. Diagnosed with nephropathic cystinosis aged 22 months. Parents first child.
- Early signs – excessive thirst and urination.
- Mum's concerns ignored by health professionals – an over anxious mother, I 'was a miserable baby.'
- 12 months old rushed to hospital – losing weight every day.
- Several months later diagnosis made, cysteamine treatment shortly before 2<sup>nd</sup> birthday.
- At diagnosis parents informed I would need a kidney transplant by age 5, would not live past 20.
- Kidney transplant aged 23 – 1<sup>st</sup> October 2009.

# Teenage Years

- No problems with appetite.
- School.
- Average age for onset of puberty.
- Developed scoliosis aged 13, surgery aged 15. Complication of cystinosis? Side effect of medication? Bad luck? Lack of research in this area.
- Transition from paediatrics to adult services difficult – did not take health seriously. Kidney function began to decline.

# Compliance with Treatment

- Pre-transplant – unwell if didn't take meds. Little side effects from cystagon, therefore compliant growing up.
  - Increased native kidneys lifespan.
  - No other complications commonly associated with adults with cystinosis e.g. diabetes, hypothyroidism, muscle wastage.
- Photophobia – mercaptamine eye drops difficult to administer regularly but used daily. Consequences if don't use eye drops – blindness.

# Cystinosis clinic



- Throughout life under care of renal department.
- Cystinosis a metabolic condition, affects the whole body, not just the kidneys.
- Attended the Cystinosis Foundation UK's conference in 2011, Dr Lipkin presented on transition from paediatrics to adult care, and cystinosis clinic.
- Attended clinic in 2012 for tests.
- Annual check ups since.

# Leading a 'normal' life



- Graduated from University of Chester in 2007 – BSc Psychology with Animal Behaviour.
- Employed full time as a Psychological Wellbeing Practitioner for Lancashire Care NHS Foundation trust – low intensity therapy for patients with common mental health problems. Promoted to locality lead for West Lancashire in May 2012, responsible for managing service.
- Outside of work – busy social life, regular exercise. Muscle strength improved in last 2 years through exercise classes e.g.pilates.
- Overall cystinosis does not stop me doing anything, lead a normal life.
- However not all patients with cystinosis are as fortunate, affects everyone differently.

# The Role of the Cystinosis Foundation UK



The Aims of the Cystinosis Foundation UK are to:

- **Provide support** to all diagnosed with cystinosis, their families and relatives.
- **Provide information** about the disorder.
- **Publish newsletters** containing relevant information for patients and supporters.
- **Organise or participate in conferences** relevant to cystinosis.
- **Support research** into the treatment of cystinosis.

# The Role of the Cystinosis Foundation UK



- Founded in 1998 by Jonathan Terry, and charity status was achieved in March 1999.
- Funding research in to the development of prodrugs at Sunderland University, to hopefully improve treatment.
- Currently organising the 8<sup>th</sup> international cystinosis congress, along with the Cystinosis Foundation of the USA, in Manchester in July 2014.



**CYSTINOSIS**  
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**Any questions?**