

European Actions
to improve the life of patients
Living With Rare Diseases

Testimony by a Mother

Lesley Greene

Co-Founder and Vice President CLIMB

Patient Advocate EURORDIS

Member of COMP at EMA

Personal Dimension: Birth

- Born 6.50 a.m
- 8th December 1978
- Apgar Score 10
- Weight 7lbs 7oz

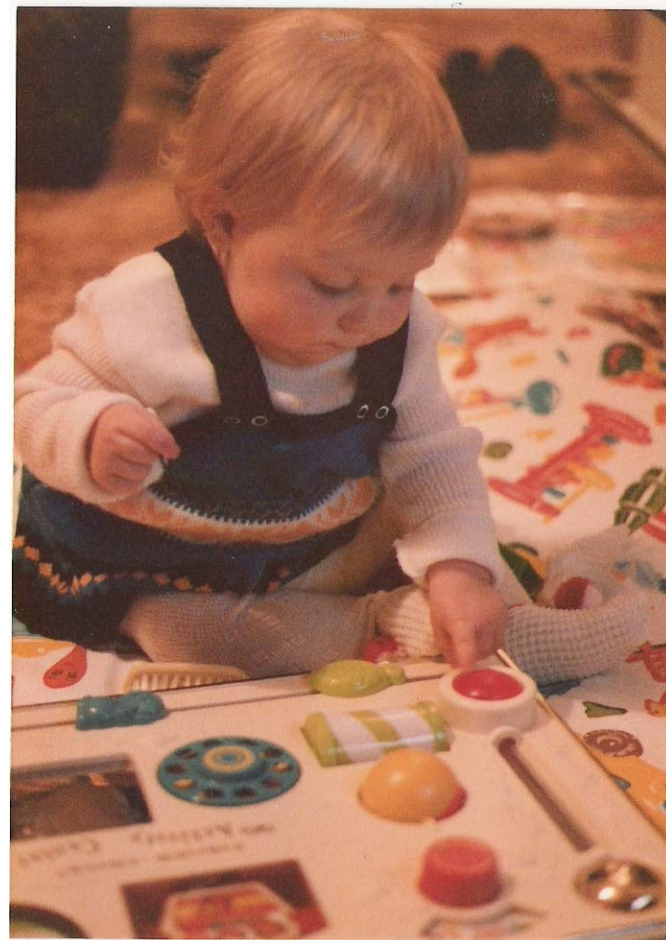


Personal Dimension: Doubts

- *Vomiting from three weeks*
- *Feeds/solids bounce back undigested*
- *Doctors and referral*

“Mrs Greene, there is nothing wrong with your baby, but we shall refer her for your sake”

- *Relief at a diagnosis*
- *New doubts*



The Personal Dimension Diagnosis of an Orphan Disease

“She has a rare, incurable, genetically inherited metabolic disease called Cystinosis which will destroy her kidney function and ultimately kill her”

Liverpool April 26th 1980



IMPACT

- My child will die. I need to know when.
- Nobody will care because it is rare
- Nobody will know how to treat her
- No-one will be doing any research
- All my babies will have this disease
- None of my family or friends will understand

Reactions



Two intertwined threads representing two intertwined dimensions-

The Life

- the medical history, physical injury, the illness, the happening, the inevitable, the unavoidable with the emotional response disbelief, grief, doubt, anger, bargaining, accepting, pain, no gains

The Emotional Cancer Journey Michele Petrone

The Mission

- the anger, the energy, the determination to act, to challenge, to hope and to change the predicted outcome



Challenges

- Rare? We will find others-in 1980 reliable data was negligible, few registries beyond PKU
- Genetic? We will find expert specialist counselling
- Untreatable? We will scour libraries, charities, researchers and expert medical contacts
- (no websites/internet, Orphanet or EURORDIS for another 17 years)

Search took Serendipity, BKPU and BBC Songs of Praise to find other “orphan parents” to support our charity mission and spe
cystinosis and beyond



Why a charity?



- **Not alone to meet the challenge; strength in collaboration and umbrella concept; more visible; more impact**
- *Driven by anger, by fear, by HOPE that our actions would make the difference: lighting the torch*

There is no medicine like HOPE, no incentive so great , and no tonic so powerful of something tomorrow

O.S Madden



1981-1991 A Life and a Charity Mission flourish

- BBC leads us to experts and clinical trial & Jen responds well to orphan drug in clinical trials
- Expert counselling, leads to birth of healthy sister
- Charity links with NORD, UK RD patient movement strengthens,
- influence on policy increases,
- funding research, more biotechs
- 1991 At 10th Anniversary Conference Charity calls for debate on Orphan drugs
- Involvement of DoH and Paediatric Medical Society; awareness of issues grows
- Orphan drugs and Medicines for Children



Momentous changes 1991-1999

- Jen and Dialysis; adjustment for all
“We have a metabolic disease”
- Transplant: flourishing growth and achievements ; gaining university entrance
- Virus leads to Burkett's Lymphoma, treatment at Centre of Expertise
- 1999 back to university and her Coming of Age





Momentous changes 1991-1999

- 1995 Charity invited to Brussels “urgent need” for Orphan Drug Regulation
- Joining forces across Europe
- Birth of EURORDIS: Orphanet
- 1999 Regulation adopted
- RTMDC comes of Age as CLIMB
(Children Living with Inherited Metabolic Diseases)



The New Millennium

- 2001 Jen receives degree
- 2001 Relapse
- 2001-2 More treatment
- 2002 Lost transplant, lost dreams (BBC)
- Dialysis
- Less choices (expertise & support as adult)
- Allowances for independence and self esteem





The new Millennium



- 2000 OD Regulation, first Patient Representatives at COMP (Committee for Orphan Medicinal Products)
- 2001-3 Elected President of EURORDIS
- 2003-8 Third Public Health Programme funded project for EURORDIS ; empowerment of patients and NGOs
- 2004 Rare Disease Task Force established & funded by PHP
- First EC funded ECRD, Luxembourg (2005) second in Lisbon (2007)
- 2007 Jen dies
- Public Health Programme funds Rapsody (improving care, information and social services for people with RD)
- 2009 EC Communication and Council Recommendation on EA in field of Rare Diseases
- 2009-present appointed member of COMP
- Europlan :empowerment of National Authorities to develop strategic RD Plans in their own countries

New torch bearers

Support from European Funded Actions continues the relay
across borders

- One life lit one torch for rare diseases
- One torch joined thousands to light up rare diseases

**Out of the darkness-
into the light**

- Together
- More visible
- More powerful
- More hope

Though we are many,
we are one voice, because we all share
one mission

To improve the lives of those living with
rare diseases

..and that is our daughter's legacy





Thank you

Additional Information

Supplementary slides

The starfish story

One day a man was walking along the beach when he noticed a boy picking something up and gently throwing it into the ocean. Approaching the boy, he asked, "What are you doing?" The youth replied, "Throwing starfish back into the ocean. The surf is up and the tide is going out. If I don't throw them back, they'll die."

"Son," the man said, "don't you realize there are miles and miles of beach and hundreds of starfish?"

You can't make a difference!"

After listening politely, the boy bent down, picked up another starfish, and threw it back into the surf. Then, smiling at the man, he said..." I made a difference for that one."

Original story by: Loren Easley



The Climb

National Information and Advice Centre for Metabolic Diseases

www.climb.org.uk: 0845 241 2173

Climb Family Services

Climb young people's service

Climb Bereavement Services

Climb Networks

Climb Conferences and Meetings

Climb Sponsored Research

Climb Support Grants

Climb Magazine and publications

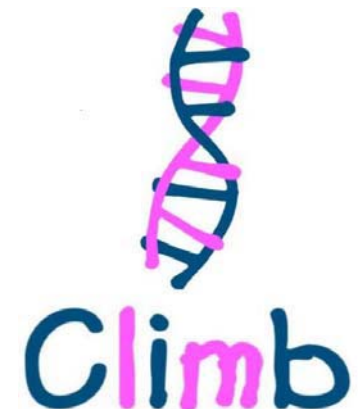
Climb web site/message forums

Climb DVDs in other languages

Climb offers and goods

National and International Advocacy and Representation

Executive Director: Mr Steve Hannigan



Cystagon and eye drops

The impact on Jen

- ✓ Cystagon-better growth, organ protection including eyes,
- ✓ Delay of renal failure=extended life-span and better quality of life
- X Number and timing of tablets
- X Nausea and odour=problems with concordance
- ✓ Eye drops; reduced photophobia and corneal erosion=better vision and quality of life
- X Maintenance and timing of eye drops
- Access

The good quality of life enabled by these treatments encourages parents to consider more children, whether affected by Cystinosis or not.

