



POMPE DISEASE

MY JOURNEY - BY SAM MURDUCK

GOSH GLYCOGEN DAY 2 FEBRUARY 2011

Background

- 1978 - Born. Normal Milestones. Active, sporty childhood
- 1992 - Teenage years – still very active (ballet, sailing) but tired all the time, persistent hip pain
- 2001 – Initial signs of muscle weakness
- 2005 – Dad died suddenly (unexplained death) – symptoms worsened around this time
- 2009 - September – Diagnosed with Limb Girdle Muscular Dystrophy
- October - Started using Bipap at night
- 2010 - February - Diagnosed with Pompe Disease
- July - Started Enzyme Replacement Therapy (Myozyme)

Signs and Symptoms

These were the problems I was experiencing for a number of years prior to diagnosis:

Frequent

- Difficulty walking up stairs and hills
- Lack of strength in legs/lower body and core muscles
- Tired all the time, no energy
- Falling asleep in the day
- Stiff/inflexible, achy, back pain crunching spine
- Tight neck and shoulders, can't bend neck forwards
- Wake up feeling slow and groggy
- 'Old lady' - take a while to get going
- Feet don't bend properly – no spring in step
- Can't take deep breaths – neck/chest too tight
- Congestion

Sometimes

- Panic attacks – mostly at night
- Purple light in front of eyes
- Shaky/restless feeling
- Headache – head exploding
- Cold hands/feet
- Numbness
- Feel spaced out
- 'Flicking' sensation in fingers
- Oversensitive to noise/smells

Rarely

- Losing vision (10mins? sweating, high temperature)
- Flashing light when eyes closed

(Many of the symptoms linked to poor respiratory function, i.e. panic attacks and morning headaches, have improved since commencing bipap).

Challenges

- **Emotional rollercoaster :**

RELIEF, FEAR, SADNESS, HELPLESS, USELESS, DEPRESSION, GUILT, ANGER, FRUSTRATION, LOSS, 'BEING A BURDEN', EMBARRASSMENT, OBSESSION, OPTIMISM, STRENGTH, POSITIVITY, FEELING LIKE A 'FRAUD', WORRY ABOUT THE FUTURE, HOPE, COMFORT

- **Feeling alone**

- **Accepting help from others**

- **My own/other peoples perceptions** – I look 'normal' but there are lots of things I find really difficult which are not obvious to others.

- **Work** – employed as an OT, need to adapt and pace myself

- **Children** – worried about genetic issues and how I would cope physically

Treatment and Support - NHS

- Neurology/Respiratory Team
- GP
- Physiotherapy
- Occupational Therapy
- Dietician
- LSDU - Enzyme Replacement Therapy
- Bipap
- Counselling – 1 year waiting list!
- Genetic testing for partner?



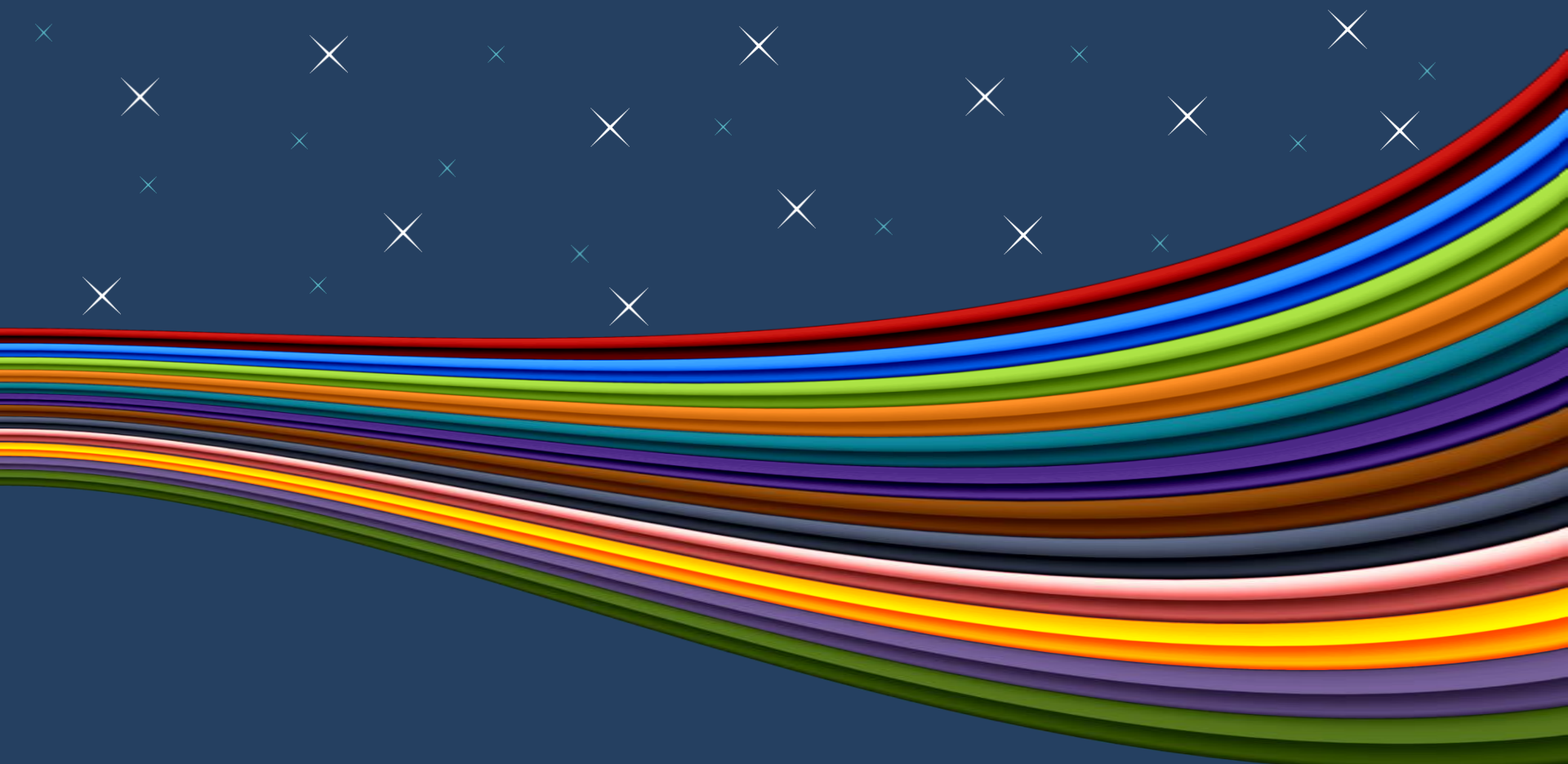
First ERT

Treatment and Support - Other



Powerplate

- Family and friends
- Exercise
- Diet – High Protein/Low Carb
- AGSD – UK
- Muscular Dystrophy Campaign
- Facebook
- GSDnet – life saver!
- Article in Pompe Bulletin
- Video interview for Southampton University medical students
- Presentation at AGSD-UK Conference 2010



POMPE POWER!!!!!!!!!!