



# PTEN Patient Day

11<sup>th</sup> November 2017



# Why are we here?

- Learn
- Make friends

# Agenda



Time	Description	Presenter
10:30	Welcome & Agenda (15 mins)	Ian Stock
10:45	Breakout sessions (45 mins)	Female, PHTS overview, Male, Parents
11:30	Support and counselling (30 mins)	Emily Pond, Katherine Lachlan and Kelly Kearley
12:00	Lunch (1 hour)	
13:00	Update on PTENUKI (30 mins)	All trustees
13:30	PTEN Research Foundation update (20 mins) PTEN Research related projects (20 mins)	Maria Whitehead Priyanka Tibarewal
14:10	A study on fat metabolism in PTEN patients (10 mins)	Professor Anna Gloyn and Dr Mahesh Umapathysivam
14:20	A study on neuropsychology in patients with PTEN alterations (10 mins)	Alice Welham
14:30	Professional Panel Q&A (30 mins)	
15:00	Coffee Break (15 mins)	
15:15	Patient registries and additional studies (30 mins)	Ian Stock and Katherine Lachlan
15:45	Patient Stories (30 mins)	Derek Small and Collette Knight

# PTENUKI UPDATE



# Why PTENUKI?

- “No work is insignificant. All labour that uplifts humanity has dignity and importance and should be undertaken with painstaking excellence.”  
— Martin Luther King Jr.
- “Do more than belong: participate. Do more than care: help. Do more than believe: practice. Do more than be fair: be kind. Do more than forgive: forget. Do more than dream: work.”  
— William Arthur Ward

# Patient Group Vision



The patient group's aim is to improve the lives of patients, parents and carers of all ages, in the United Kingdom and Ireland,

who are affected by PTEN genetic alterations, PTEN Hamartoma Tumour Syndrome (PHTS), Cowden Syndrome (CS) or Bannayan-Riley-Ruvalcaba Syndrome (BRRS)

through better **patient support**, increased awareness, more accurate and accessible information, earlier diagnosis and intervention, greater **research** into treatment and prevention, and improved coordination of **healthcare**.



# PTENUKI Update

- Charity Status -
  - What is PTENUKI? - <http://ptenuki.org/about-ptenuki/>
  - Who are we? - the Trustees
- New Website launch
- Fundraising update
- Volunteers & Contributors



# Ellie Collins (TRUSTEE - PROMOTION & AWARENESS)



- February 2014 – diagnosed just me and my Dad
- November 2017 – network of >300 people
- Helped me accept my condition
- Inspire my friends and family by talking openly about my condition
- Make friends like you!
- Rare doesn't need to be lonely...
- *Join us – let's all make one connection today!*



Private



Public





# Patient Registries

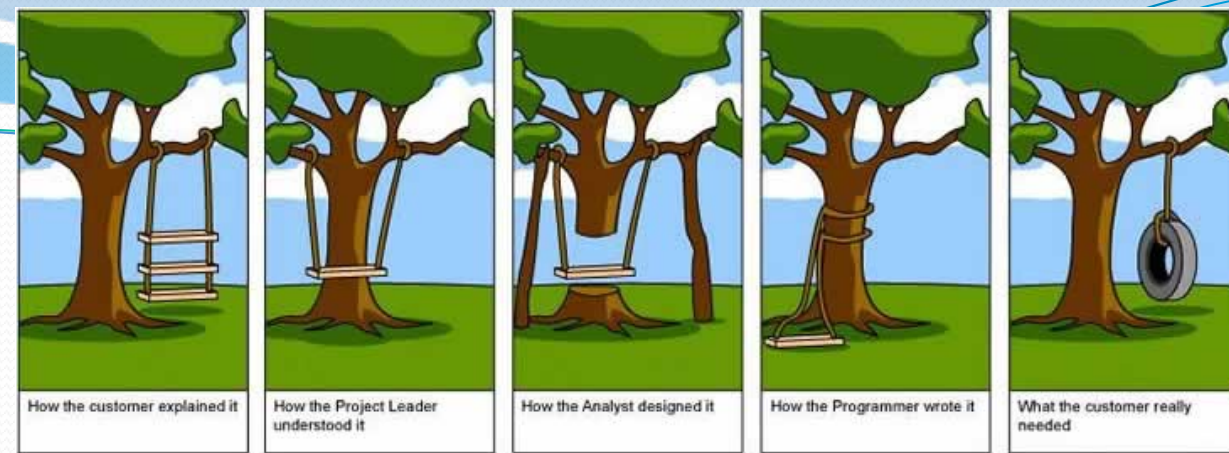
# Patient Registries (1)



- Patient registries are systems of data collection of patients with the same disease(s) from a geographically defined population over an extended period of time.
- Example data collected
  - Personal data (name, address, contact preferences, socio-demographic data...)
  - Genetic data , clinical data, family medical history
  - Medications, devices and health services used by the patient
  - Patient reported outcomes (e.g. quality of life data, health status, etc)
  - Patient participation to clinical research and bio specimen donation
- Benefits can include
  - Important tools in support of clinical innovation and improvements in care
  - Providing essential data for research, drug development and building trials
  - Help people and their families understand the disease and make informed choices
  - There is evidence that quality of care and life expectancy improves dramatically.

# Patient Registries (2)

- Patient groups want and are capable of being involved in registry development. They can contribute to :
  - the selection of data items collected
  - initiating the establishment of registries by creating partnerships with professionals
  - collecting data (in particular about the impact of the diseases on their daily life)
  - helping recruit patients to participate
  - helping recruit interested health professionals to input data
- Patients should be involved in development, management and maintenance to:
  - best represent patient needs
  - increase awareness among all
  - Be stakeholders of the existence of the registry
  - ultimately improving the quality and quantity of data collected
- So what are the next steps...?



# Patient Registries (3)

- Flóra Raffai, Findacure: <https://goo.gl/C5J1oJ>
- Chris Sotirelis, RDConnect: <https://goo.gl/oIMvCM>
- Avril Kennan, DEBRA Ireland: <https://goo.gl/xjph9N>
- Rebecca Cosgriff, Cystic Fibrosis Trust: <https://goo.gl/B4xVCz>
- Asha Hareendran, Evidera: <https://goo.gl/fH5SUZ>



- Experiences and Challenges
- Benefits and Recommendations
- Funding & Solution options
- Patient involvement



