Patient voices in Personalised Medicine

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PART 1
Sam's story

A brief history

- At birth: perfect second child
- By 3 months: severe visual impairment
- By 1st birthday: global developmental delay
- By 2nd birthday: status epilepticus
- Today (8yrs): non verbal, not walking, registered blind, spoon fed, multiple daily medications

Our ‘diagnostic odyssey’

- Enigma: many invasive tests but all results “normal”
- Blood samples taken for trio WGS (Sept 2015)
- Genomics England found 43 genes of interest
- Further reading pointed to GRIN1 gene
- Local NHS check confirmed this very likely, but asked for our opinion as well
- First ever NHS patient to be diagnosed with GRIN1 disorder
- De novo mutation

Impact of diagnosis

- No prognosis, but…
- Found a community around the world
- Helped to set up a non-profit organization in search of treatments and cures (CureGRIN.org)
- Community growing all the time: GRIN1 gene now on panel tests for intellectual disability & epilepsy

Lessons learned

Patients and their families hold pieces of the jigsaw that can help to find new diagnoses

Never assume that a family doesn't have the capacity to understand. Parents are among the most motivated people in the world to learn new stuff when it affects their children, and to drive progress forward

Consent requires the patient/family to understand what is on offer, what genomics can and cannot (currently) do. Analogies can be useful (e.g. encyclopaedia, beads on a necklace, windows on a train)