A GP’s journey with a rare disease

Dr Annette Scivier

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“Rare diseases”

- Defined as a condition that affects less than 5 in 10,000 or 0.05% of people in the population
- Often rare diseases are chronic and life-threatening
- Rare diseases can be single gene, multifactorial, chromosomal or non-genetic
- There are over 6,000 recognised rare diseases
- **Collectively rare diseases are not rare….**
  - 7% of people will be affected by a rare disease at some point in their life

  (Diabetes affects ~6%,not all diagnosed)
  - Approximately 3.5 million people in the UK

  (Diabetes affects 3.5 million)
  - Neurological conditions affecting adults and genetic diseases form a large proportion of these rare diseases

Long-term neurological conditions

Long-term neurological conditions (LTNC) refer to conditions where damage to the nervous system results in impairment to normal functioning over a period of time.

- Common conditions:
  - Stroke
  - Epilepsy
  - Multiple sclerosis
  - Parkinson's, Alzheimer's etc
- Rare neurological conditions:
  - Charcot-Marie Tooth
  - Motor neurone disease
  - Huntington's disease
  - Progressive supranuclear palsy
  - Multiple system atrophy

Table 4.4 Estimates of disease incidence and prevalence from THIN database

	Year				
	2004	2005	2006	2007	2008
Population denominator	2,840,202	2,948,206	2,977,044	2,983,870	2,964,386
Condition					
Motor neuron disease					
Incidence	3.24	3.53	2.55	3.38	3.14
Prevalence	9.99	9.91	9.37	10.08	9.75
Huntington's disease					
Incidence	0.70	0.78	0.64	0.50	0.44
Prevalence	6.45	6.51	6.54	6.52	5.96
Charcot-Marie-Tooth					
Incidence	1.48	2.17	1.88	2.14	1.52
Prevalence	8.42	10.01	11.56	12.90	12.79
Progressive supranuclear palsy					
Incidence	0.60	0.54	0.81	0.77	1.21
Prevalence	1.70	1.86	2.06	2.09	2.34
Multiple system atrophy					
Incidence	0.32	0.54	0.44	0.44	0.37
Prevalence	0.68	1.01	1.13	1.19	1.12

Note: Incidence (per 100,000 person years); Prevalence (per 100,000 of population)

Lack of research of these conditions means difficulty in recognising them, lack of data on life expectancy and lack of understanding of disability impact

Source: http://www.ltnc.org.uk

The start of my journey

- Met when I was age 18, Husband was 22, married 5 years later
- Told that his father had died, 10 years after his parents divorced
- His father had become “depressed” and lived at a Psychiatric Hospital until a sudden death when my husband was 18 , having not seen him for about 9 years.
- His father was 49

The road gets tough…

- Age 32 my husband leaves his Naval Officer career and completes an MSc, whilst I get a new job
- He leaves 4 jobs in 4 years (and got a new mortgage during 3rd job)
- Our 1st child arrives according to plan - I return to work to pay the bills, we don't get round to Wills and Life Insurance

…and tougher

- Relationship was strained next 2-3years
- “normal” tiredness and “male” behaviour around the house – eg multitasking, chores
- Washing up skills lacking
- Many DIY projects started, not finished
- Mood swings evident between jobs

…ups and downs en route

- Mood swings became aggressive with suicidal threats
- Scary
- Attributed to his job not being the right one, difficult commute, or….lack of job
- Refused to get help…. his medical record may affect job hunting
- Refused to apply for benefits…I had a job

The journey continues…

- I had by now received counselling from 3 organisations
- Our 2nd daughter was born and I returned to work ,my job itself becoming very stressful
- Thankfully we left a crumbling cottage at his next job change
- Left that job twice, half completing a teacher training course three times
- During this period friends and family began to become concerned about what was wrong- was it depression?
- Still refused to accept any suggestion to see a GP except for some urine infections/ symptoms of renal stones
- A Will was made for the first time and quotes obtained for Life Insurance

Diagnosis by Google….

- NOT to be recommended for Huntington's Disease
- NOT what my best friend intended when she said I really needed to find out why my husbands behaviour and movements were odd for 10 years
- Reading the leaflet "behaviour problems" on Huntington's Disease Association (HDA) charity website it suddenly "clicked" in my head what I had been living with
- HDA website:

  "The symptoms of Huntington's disease usually develop when people are between 30-50 years old, although they can start much earlier or much later. The symptoms can also differ from person to person, even in the same family."

  "Sometimes, the symptoms are present for a long time before a diagnosis of Huntington's disease is made. This is <u>especially true when people are not aware</u> that Huntington's disease is in their family."

Diagnosis by Google….

Classic symptoms from HD website information:

“The early symptoms include:

- slight, uncontrollable muscular movements
- stumbling and clumsiness
- lack of concentration
- short-term memory lapses
- depression
- changes of mood, sometimes including aggressive or antisocial behaviour”

Source: www.hda.org.uk

The hurdles of the NHS begin ….

- His kind GP accepted my word on the history without his patient attending and phones a neurologist (*unheard of !)*
- Seeing the patient helps with diagnosis of movement disorder…not easy if you don't believe you are ill
- Letters and phone calls from neurology secretary are misunderstood by the patient (assumed to have intact capacity and organisational skills)
- Bike accident needing stitches convinces him and his family that, yes, something is odd about his balance and twitching
- Spurious blip on an ECG in casualty convinces my husband that return visits to his GP are needed

Lack of signposts for professionals

- My husband managed half a neurology outpatient appointment - long enough to confirm my suspicions
- “Open appointment” given to return – DNAx2
- Neurologist established that I had been in touch with HDA –I felt guilty knowing Regional Care Advisor covered 3 counties
- No nurse in clinic to ask if I had any questions or needed information
- No suggestions to his GP on who was available locally, or elsewhere, with experience of HD

Help & Support?

Who does what in a challenging illness?

Who guides the way?

Who advises when an illness blows your family apart?

- Regional genetics service, Southampton
- National Research team at Queen Square, London
  - Refusals to see adult psychiatrist locally
  - Agrees to speech therapist + dietician but not local neurologist or physiotherapist

Navigating the maze of a critical illness

- A fatal, rare (or not so rare) critical illness prevented gainful employment for 3-10 years before diagnosis, then that of a partner for an indefinite period due to first stress then caring responsibilities

- During prolonged " crisis" due to challenging behaviour of early dementia resulted in 18 months of emotional and financial uncertainty as long term care was suggested and eventually found

- Being in limbo I sought advise from 57 professionals in medical, social, housing ,legal and financial realms

- As a new self employed partner I had taken out a locum policy, then PHI policy on which we have depended

Gaps in services

- Neurology or psychiatry
- Adult psychiatry or old age psychiatry – depends where you live
- Young Carers between contracts -6m gap
- Emotional support for me (main carer)-unclear
- Carers assessment – 5 phone calls
- Websites and services (eg advocacy and mediation) for dementia and mental health do not consider HD to be part of their remit
- Mental Health Act or Family Law Act?
- NHS England is unsure if neurology services are commissioned locally by CCGs or by national Specialist Commissioning

Coordination of rare conditions

- Medicine is pattern recognition
- I Googled movement disorder to make a diagnosis – NOT recommended!
- Confirmation can easily take months or years
- Targets do not exist for many specialities
- Data does not exist
- Funding very limited
  - 2008/09 the rare diseases received the equivalent of **£1 of charitable funding per patient**, compared to **£185 per patient for cancer**

HD – what we know now…

Estimated prevalence:

- 1990 estimate = **5.4** per 100,000 (95% CI 3.8 to 7.5)
- 2010 estimate = **12.3** per 100,000 (95% CI 11.2 to 13.5)
- Age specific prevalence was highest in the 51-60 year age range (15.8 per 100,000)
- Increase in prevalence due to more accurate diagnoses, better and more available therapies and an improved life expectancy.
- Greater willingness to register a diagnosis of HD in patients' electronic medical records.

Prognosis previously under-estimated?

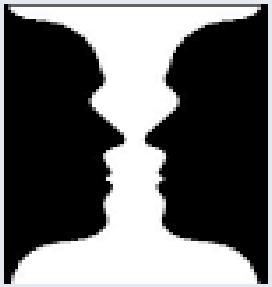
- Mean age at onset presumed 30 to 50 but LTNC study (2011) suggested mean age 48
- Survival from diagnosis 15 – 20 years; LTNC study found median survival 38 years

Difference may be due to taking survival from date of pre-symptomatic genetic test rather than diagnosis after onset of symptoms…

Treatment and management do make a difference

- Maintain high calorie intake 5-7,000 calories daily
- Regular physical exercise – good evidence
- Better understanding of neuropsychology and behavioural management , input from specialist rehabilitation
- Recent years of well funded (USA) focused trials – will make a difference to my children's future since accurate gene tests in 1993. My husband is amongst thousands on a secure database waiting to contribute to research.

A neurological condition or just “normal life”?



- Underwriters and Claims Assessors get to see the history over a long time; patterns might be easier to spot than in a 10 minute appointment with a GP
- Looking backwards you may see the clues that a GP, the patient/customer or their family had not spotted

Balance of assessment:

- Distress if “label” is wrong e.g. unnecessary exclusions on insurance policies
- Identifying a life-threatening condition could help with instigation of support and treatment

HDA advice:

“Some people who know they are at risk spend time searching for the first signs that they are developing the disease.

They may worry about simple things like dropping a cup, forgetting a name or becoming unusually bad-tempered. ***Most people do these things occasionally*** - whether they are at risk from Huntington’s disease or not - so they could be worrying unnecessarily”

If you have a rare illness you expect the same access to financial advice, NHS treatment and may have even more need of critical illness and life insurance

Treatment and management do make a difference

Public Accounts Committee Report on Services to people with neurological conditions: a progress review
Feb 26th 2016

"wide variation across the country in access, outcomes and patient experience" and in particular that "diagnosing neurological conditions takes too long, services in hospitals are variable and local health and social care services are often poorly coordinated".

"Our Committee heard of unsettling disparities in outcomes for people living with what can be devastating or even fatal conditions. We also heard of the huge knock-on costs the NHS can incur as a result of poor care."

This Report serves as a wake-up call to the Department of Health and NHS England. The message from the frontline is clear and central government must listen and learn.

Questions & more information

http://hda.org.uk/

Huntington's Disease Association
England & Wales
Hunting for a cure, with care

http://en.hdbuzz.net/

HDBuzz

http://www.neural.org.uk/

the neurological alliance