Huntington disease

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J R Soc Med 2005;**98**:550

Huntington disease affects between 6500 and 8000 people in the UK and has a worldwide prevalence of about 1 in 20 000. It is a genetic disorder of the central nervous system with symptoms usually appearing in the third or fourth decade of life. It shows autosomal dominance with full penetration and is caused by a single defective gene on chromosome 4. This has been attributed to expansion of glutamine repeats which leads to neural death in the basal ganglia and cerebral cortex and to the gradual onset of physical, mental and emotional changes.[1] These include involuntary chorea form movements, difficulty speaking, depression, anorexia and loss of rational thought. The disease may progress slowly but it is an accepted tragedy that, by the time the symptoms first appear, the person has had their own family and possibly already passed on the genetic problem.

George S Huntington (1850–1916) was an extremely ambitious but newly qualified young doctor when he set out on rounds with his father, also a physician, in Pomeroy, Ohio in the second half of the nineteenth century.[2] It was the sight of 'two women, mother and daughter' which inspired a lifelong interest in a disease which had already been 'discovered'! Johan Christian Lund (1830–1906) was an unassuming Norwegian who made the first recorded description after working near Setesdal. His paper was published in 1860 when George was nine.[3] In it he describes the classic pattern of symptoms and its obvious hereditary nature. However, it was not translated into English until 1959. Meanwhile, Huntington published his discovery at the age of 22 to great critical acclaim—and shortly afterwards the eponym was being used throughout the USA and Europe.

I suggest that as this disease was originally recognized by Lund we use his modest and more European name setesdalryyja, rather than concentrating on Huntington's Career.

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REFERENCES

1 Perutz MF, Windle AH. Cause of neural death in neurodegenerative disease attributable to expansion of glutamine repeats. *Nature* 2001; **412**:143–4

2 Huntington G. On chorea. *Med Surg Reporter Philadelphia* 1872;**15**: 317–21

3 Lund JC. Chorea Sti Viti I Stersdalen. *Uddrag af Distriktsoege J C Lunds Medicinalberetning for 1860*. [Beretning om Sundhedstilstandenm.m. i Norge i 1860: 137–8. English trans. 1959]