Huntington’s disease (HD) is a progressive neurological disorder characterized by involuntary movements, cognitive dysfunction, behavioural changes and psychiatric illness. HD is a rare genetic disorder; the prevalence rates vary worldwide, but in Australia, North America and Europe it has been estimated that 5.70 people are affected per 100,000 of the population (Pringsheim et al., 2012). George Huntington, a medical doctor, first described HD in 1872. He described it as “hereditary chorea”, chorea meaning “dance” in Greek and referring to the involuntary movements so common in this disease, which are reminiscent of dance movements (Huntington, 2003).

**Genetics**

HD is an autosomal dominant genetic disease, which means that only one parent needs to have the gene to pass it onto their child, and the child of an affected parent will have a 50% chance of inheriting the disease. The gene mutation responsible is an unstable enlargement of a CAG repeat sequence which produces a protein called huntingtin. This Huntingtin gene was identified in 1983, and since then a predictive test has been developed which allows individuals to determine if they carry the gene (Adam & Jankovic, 2008). While the disease age of onset varies from person to person, initial symptoms are most commonly detected between the ages of 35 and 55, although juvenile or late onset does occur (Forrest Keenan, Simpson, Miedzybrodzka, Alexander, & Semper, 2013). The extent of the genetic mutation is generally related to the age of onset, with larger numbers of CAG repeats associated with earlier onset. Because the symptoms generally emerge in the middle adult years, affected individuals often have children before their diagnosis, and so their children are at risk. While many individuals at risk now seek out predictive testing, predictive testing often has potential implications for an at-risk individual and their family to consider. Genetic counselling is therefore recommended before making this decision (Forrest Keenan et al., 2013).

**Symptoms**

The first area of the brain to be affected by HD is the “striatum”, which is important for learning, movement, and other cognitive functions (Paulsen et al., 2006). The disease targets particular brain cells (neurons) in the striatum, which are important for controlling movement. Therefore changes to movement, including chorea, rigidity, and motor slowing are common. In the addition, the striatum is important for particular cognitive abilities, regulating mood and controlling behaviour. With regard to cognition, patients with HD often show impairments in their mental speed, attention, planning, judgement and reasoning, as well as inefficient memory (Papoutsi, Labuschagne, Tabrizi, & Stout, 2014). In addition, behavioural issues such as irritability, apathy, and aggression are common. Psychiatric
illness, such as depression, anxiety and occasionally psychosis, can occur. The combination of symptoms and the severity of those symptoms vary substantially from patient to patient. While the symptoms are often initially mild, they generally steadily worsen over time and usually cause disability and death within 15-20 years (Dorsey et al., 2013; Frank, 2014).

**Treatment**

Unfortunately there is not yet a treatment for HD that can reverse or halt the progression of the disease. At the current time HD treatments are primarily pharmacological and focused on managing the symptoms of the disease, that is, medications targeting the motor, behavioural and psychiatric symptoms, which include neuroleptics and anti-depressants (Frank, 2014). There is not yet an effective treatment for the cognitive symptoms of the disease. Invasive treatments such as deep brain stimulation and fetal cell transplantation have been trialled in small numbers of patients with mixed results, and these techniques are not used in usual clinical practice (Adam & Jankovic, 2008). There are some early indications that the onset of the HD and its progression might be delayed or slowed by certain environmental and lifestyle factors, such as physical exercise, education and employment, however more research is needed to understand this better (Bonner-Jackson et al., 2013). Management of patients’ needs generally involves a multidisciplinary approach, which can include input from a neurologist, psychiatrist, occupational therapy, social work and palliative care, among others.

**References:**


