Huntington’s disease (HD) is an inherited disease which causes progressive damage to the brain. The disease is genetic, and has been traced back to a single gene which can be detected using blood tests. This gene causes an extended DNA chain of chemicals to occur within a protein called Huntingtin (Htt). This chain is called the cytosine-adenine-guanine (CAG) repeat, named after the particular chemicals involved. Though its functions are not completely known the Huntingtin protein plays a role in the natural death of cells within the body. When an individual has over 39 CAG repeats the Huntingtin protein becomes unstable. As a result, the protein behaves abnormally in the brain and forms clusters, causing premature death of brain tissue both across the brain and within specific structures. Brain damage begins up to 15 years before an individual develops symptoms of the disease, occurring at approximately 40 years of age.

Genetic testing is used to count the number of CAG repeats present in an individual’s Huntingtin gene, determining if someone will develop the disease later in life.

The disease is typically viewed as having two main stages. There is the early, dormant phase of the disease, where the individual does not experience any symptoms. This occurs approximately between birth and age 40. The second stage is termed the symptomatic stage. This occurs as the particular patterns of damage affect the circuitry in the brain which controls movement, mood, and other brain functions. Motor symptoms involve both impairment to existing movements as well as the addition of uncontrollable movements. For example, individuals typically struggle to walk, utilize cutlery, and feed themselves. They also may develop a tremor, or shaking, dance-like movements (termed chorea). Mood disturbances also can occur, including increased aggression, personality changes, depression, and psychotic features. Damage to brain circuitry impacts on the individuals’ mental functioning. Memories, organization and planning abilities all decline. Symptoms develop over a course of 15-20 years, with individuals progressively losing their ability to walk, talk, and feed themselves. They will become incontinent, are often confined to a wheelchair and will likely be placed in palliative care. Development of dementia is typically seen in late stages of the disease. Death occurs from secondary symptoms of the disease, such as complications from falls, infection, or choking on food (aspiration pneumonia).

Due to its nature, Huntington’s disease (HD) often involves multiple medical and health professionals in caring and providing support for individuals. These frequently include a medical specialist called a Neurologist, who can assess whether a patient has motor symptoms, and a genetics counselor, who may provide genetic testing and advice regarding results. In later stages, speech pathologists and occupational therapists may assist individuals with progressive swallowing and movement difficulties. Family counseling is often important due to the genetic nature of the disease and the difficulties faced by family members in caring for a relative whilst considering their own genetic testing.
There are no known cures or preventative treatments for Huntington’s Disease. Currently, some symptomatic treatments are used to treat particular symptoms, such as psychotic episodes and tremor. However, these do not influence the rapid progression of the disease. While individuals do not develop symptoms of the disease until later in life, studies using brain imaging have identified that the Huntington’s brain begins to change up to 15 years prior to symptoms. These changes suggest that the brain begins to experience damage due to the abnormal huntingtin protein, which is thought to build up in the brain over the lifespan of an individual. Neural changes include loss of brain volume or shrinkage, which is commonly termed atrophy. Evidence of brain changes prior to Huntington’s symptoms has given support to a worldwide research effort to identify early signs of HD. It is important to discover these changes so that preventative treatments may be trialed in patients and so that we can understand whether these treatments are effectively preventing the disease from progressing to later, aggressive stages.