Huntington's Chorea in South Africa

A national investigation into the origin and frequency of Huntington's chorea in all population groups of South Africa has been carried out by Dr M.R. Hayden and colleagues of the Department of Human Genetics, University of Cape Town. A total of 481 persons, of whom 153 are still living, has been recorded. The disease is a lethal genetic neuro-psychiatric disorder characterised by chorea and dementia. Symptoms generally appear in adulthood and death usually occurs within 10 to 15 years of their appearance.

The prevalence of the disorder of 22,2 per million in the White (mainly Afrikaner) and 21,7 in the Coloured groups is more than 200 times greater than the calculated frequency of 0,1 in Blacks. The most likely explanation for this finding is that the gene for the disease in South Africa has its origins in north-western Europe. In view of the fact that there has been relatively little genetic drift between those of European descent and the African negro group, it is likely that the gene would be uncommon in the latter population. The low prevalence in the African negro is the lowest reported figure in the world.

Over 200 affected individuals in more than 50 supposedly unrelated families (constituting 80 per cent of all the Afrikaner patients ascertained during the survey) have been found to be ancestrally related through a common progenitor who came to the Cape of Good Hope as a free burgher in the 17th century Willem Schalk van der Merwe, who came from near Rotterdam in Holland, was among the 344 persons who arrived on the *Dordrecht* at the Cape in 1658. In 1668 he married Elsjie Cloeten, whose father had arrived at the Cape with Jan van Riebeeck in 1652. Their first child, Sophia, married Roelof Pasman in 1684, by whom she had five children. In 1696, after Pasman's death, Sophia married Pieter Robberts. A total of 210 affected individuals have been traced over 14 generations from the present day to the two marriages of Sophia van der Merwe. It is thus probable that she carried the gene for Huntington's chorea.

South Africa offers unique opportunities for genealogical projects in view of the excellent recording of the origins and history of the Afrikaner nation, mainly in the archives of the Dutch Reformed Church. These documents facilitated the medical team's efforts to trace the earliest transmitters of the gene to this country. Apart from personal visits to the homes of the affected person's parents to obtain genealogical information, church archives, entries in family Bibles, old letters and even gravestones were examined in a bid to trace the kinship's history.

Ignorance of the disorder, its mode of inheritance and implications, among the public and the medical profession, has resulted in a high proportion of misdiagnoses (approximately 40 per cent in this survey). Such poor awareness of the disorder has added to the existing social stigma and superstition attached to the disease, with patients being reluctant to present themselves for diagnosis.

Hungtington's chorea should be suspected in patients who present with unexplained psychiatric or neurological symptoms, including personality change, anti-social behaviour or abnormal physical movements. In these instances, a family history of the disorder should be carefully looked for, particularly in areas such as the north-western Cape, Where there is a concentration of affected families.

The social implications of Huntington's chorea are serious and far-reaching, all members of the family and the community as a whole. The relatively common occurrence of suicide and of major and minor crimes by those affected are cause for concern. The disease imposes a significant economic burden on both the family and society. The minimum direct cost of treatment to the State of a single person affected with Huntington's chorea in South Africa is estimated at R23 000. Every child of an affected parent has an even chance of inheriting the gene. At present a childless marriage by those at risk is the only way to decrease the incidence of the disorder.

While there is as yet no medical cure available, the major task of persons involved in the management of affected patients and their families is to provide genetic counselling and improved care. Appropriate pharmacology can alleviate many of the patient's symptoms. With greater understanding of the disease, the social stigma will diminish and families will feel free to use the available community resources, such as the special Huntington's chorea clinic which has been established at Groote Schuur Hospital.

Awareness of the need for comprehensive care for the whole family in cases of Huntington's chorea highlights the unmet needs and socio-economic implications of persons suffering from numerous other unrelated genetic and chronic disorders.

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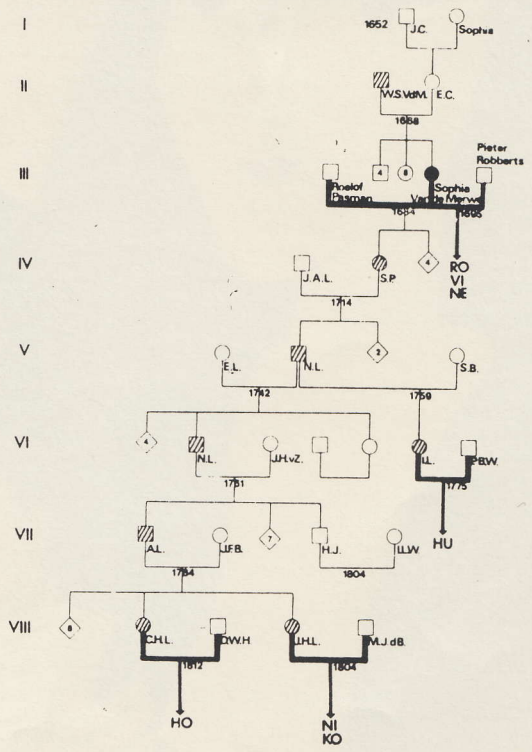

Pedigree showing the relationship of over 210 affected persons to their common ancestor in the 17th century.