Pendred’s Syndrome

J.M.S. Pearce
Emeritus Consultant Neurologist, Department of Neurology, Hull Royal Infirmary and Hull York Medical School, Hull, UK

Key Words
Pendred’s syndrome · Sensorineural deafness · Goitre

Abstract
Vaughan Pendred, whilst still a junior doctor, wrote the original account of familial sensorineural deafness associated with goitre, now known as Pendred’s syndrome. His account is reproduced verbatim.

Neurologists sometimes encounter Pendred’s syndrome, one of the most frequent causes of congenital deafness, accounting for about 10% of all cases of hereditary deafness. Pendred (1869–1946) was a Guy’s Hospital graduate and is said to have become a general practi-
years, and the mother, aged sixty-seven years, are alive and healthy. They have had ten children, five sons and five daughters. In an episode of small-pox twenty-five years ago the whole family was attacked with the exception of the younger of the deaf-mutes, and four males and one female died, although all had been vaccinated, and this recently, as they were children. The remaining son and two of the daughters are healthy and vigorous.

The first goitre case is the first-born of the family—a spare woman now aged thirty-eight years. She is deaf and can only mumble indistinctly; little care has been taken to educate her and so she is imbecile. The goitre is a large multilobular hard tumour, the greater part on the right side of the neck; from time to time she suffers from dyspnoeic attacks. The growth was first observed after the small-pox—i.e. at thirteen years of age. The second surviving girl is now aged twenty-eight years and is the fifth of the family; she is a small, spare, intelligent woman, her expression being in marked contrast to her sister’s. She is not absolutely deaf and can mumble incoherently; her education has been attended to with so much success that she has been ‘in service’. The tumour is larger than in the other case, but is of the same character; it has been growing for about fifteen years, and during the last year has caused both dyspnoea and dysphagia, which have become so urgent that I have sent her to-day to Newcastle Infirmary for operation.

D u r h a m .

It was exactly a century later that Coyle et al. [3] and Sheffield et al. [4] in consecutive papers in *Nature Genetics* showed that the disorder maps to chromosome 7q.

References


