

SORSBY'S FUNDUS DYSTROPHY. Sam Hamilton.

Margaret and Dixon Scorer examine the yellowing scroll of paper depicting Margaret's family tree. "Where's Joseph Park?" asks Margaret, 69. Dixon points out his wife's great grandfather from whom the six generations depicted are descended. This pedigree uses solid black symbols to denote blind family members and open symbols for the sighted. Margaret, herself registered blind 30 years ago, commissioned it back then. The number of solid black symbols added weight to her suspicions that her blindness was inherited.

"It just happened overnight" said Margaret. "One morning in 1963, I awoke to see bulges in the door jambs and window frames. My sight deteriorated in one eye only at first, but eighteen months later, my other eye became affected and within four weeks, I had no central vision in either eye". Margaret, determined not to beaten, underwent rehabilitation, and subsequently worked as an audio typist for 16 years, ran a home and brought up two young children. This remarkable woman sees nothing special in her achievements. "I just got on with life as best I could. I didn't want the family to see how it was affecting me in case one day the children developed the same thing" she said.

Margaret's condition could not be confirmed until her pedigree came to light in 1990. The discovery that Margaret was descended from the Park family, enabled Sorsby's fundus dystrophy, SFD, to be diagnosed. The disease, named after its discoverer in 1949, is a very rare form of inherited macular dystrophy, causing irreversible loss of central vision in 20 to 40 year olds, often beginning with deterioration of night vision. Commoner forms of macular disease are responsible for half the world's blindness. Although the worldwide incidence of SFD is unknown, a pocket of three affected families in the North East, contributes to a higher than predicted incidence of the disease here than elsewhere in Britain, placing Newcastle University staff at a research advantage.

SFD is distinguishable from other macular disease by its onset age and characteristic changes in the retina which cause the blindness. Electrophysiological data, generated by the Medical Physics department, plays a crucial part in diagnosis. The patient is placed in a darkened room with electrodes connected to their temples and in the lower eyelid of the affected eye. Lights flashed at the subject result in electrical responses from the retina which are recorded via the electrodes. These responses are affected in a particular way in SFD.

Sharon McDonnell, the current genetic liaison nurse for ophthalmology, is tracing possible North East Sorsby's sufferers through pedigrees like Margaret's. Genetic principles explain why only some members of Sorsby's families inherit SFD. We inherit our characteristics from our parents as a set of "genetic commands" or chromosomes from each of them. Chromosomes, in all our body cells, are made up of many smaller elements called genes. Our genes determine all our characteristics, for example, eye and hair colour. They also determine whether or not we may go on to develop certain diseases, including SFD. The child of a Sorsby's sufferer (who has the Sorsby's gene) and a normal individual (who does not have this faulty gene) will have a 50:50 chance of developing Sorsby's later in life. Only one of Margaret's two children has developed SFD in recent years. The other has not and a recent blood test found her to be clear of the faulty gene. Tragically, before the advent of genetic testing, the affected parent would not usually develop the disease until well after they had had their children and would be unaware that they could be passing on a form of inherited blindness to the next generation.

In 1993, German scientists identified the Sorsby's gene. Later, they realised that another gene, crucial in controlling levels of eye protein and closely positioned to the Sorsby's gene, was faulty only in Sorsby's patients, but not unaffected relatives. The resulting changes in eye protein could cause visual damage, indicating Sorsby's was a result of this genetic defect. Now, couples could, theoretically, be tested before starting a family, to determine if they are SFD carriers, although the test is not yet widely available. It could help couples to make informed decisions when planning a family and may contribute to a reduction in suffering. Research for those already suffering from SFD is also taking place. In late 1995, an American group discovered that in the early stages of Sorsby's, the night blindness often experienced is due to the fact that dietary vitamin A, essential for good night vision, cannot reach its target in the eye because of the abnormal levels of eye protein. Massive doses of vitamin A given to early stage patients suffering night blindness, actually reversed the blindness within a week and kept it away as long as the patient took the vitamin A. Although the long term safety and effectiveness of such treatment has yet to be tested here in the UK, in future, vitamin A treatment may help to control the early stages of blindness. With larger numbers of potential SFD sufferers in the North East than elsewhere in the UK, trials for early stage sufferers are a long term aim for Sharon and her colleagues, alongside the genetic counselling currently undertaken.

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