Joseph Cary Merrick, the most famous case-report of neurofibromatosis type I

Many observations of what was soon to be called neurofibromatosis have been published in medical literature before the description of this disease by the German professor of pathology Friedrich Daniel von Recklinghausen in 1882. Some of them showed impressive neurofibromas (Virchow, 1863-1867), but Joseph Carey Merrick, otherwise known as the Elephant Man, unquestionably deserves to be considered as the most famous case-report of neurofibromatosis.

Neurofibromatosis
Neurofibromatosis belongs to phacomatoses, a group of congenital neuroectodermal dysplasias including tuberous sclerosis (described by Désiré Magloire Bourneville in 1880) and angiomatosis retinae (described by Eugen von Hippel in 1895 and Arvid Vilhelm Lindau in 1926). Neurofibromatosis type 1 (Recklinghausen’s disease) has the following major features: multiple neural tumors (neurofibromas), pigmented skin lesions (including café au lait spots), skeletal lesions (scoliosis, pseudarthrosis of the tibia), and pigmented iris hamartomas (Lisch nodules) (Cabanne et Bonenfant, 1986; Kumar et al., 2005). Neurofibromatosis type 2, previously called acoustic neurofibromatosis, is another disorder which has no link with the present column.

Joseph Carey Merrick
Joseph Carey Merrick, the first son of the storekeeper Joseph Rockley Merrick and Mary Jane Potterton, was born at Leicester on August 5, 1862. About three months before his birth, his mother narrowly missed to be crushed by an elephant of the George Wombell travelling menagerie at Humberstonegate. That’s why Joseph Carey has been remembered as the Elephant Man. In his first year of life, the child seemed to be totally normal, but malformations began to appear around the age of 21 months: a firm mass of the lower lip extended to the right cheek, the skull developed a large frontal protuberance, both feet and the right upper limb enormously enlarged. His skull perimeter reached 90 centimeters, and that of his right wrist 30 centimeters.

Joseph’s mother died of pneumonia on May 19, 1873. Eighteen months later, on December 3, 1874 his father married again Emma Wood Antill who soon managed to get rid of the poor boy. Joseph Carey had therefore to hold some jobs – working man in the Freeman cigars factory at Lower Hill Street, gloves and stocking door to door travelling salesman -, but his ever growing deformities made him soon out of work. He then lived for a time by his uncle Charles Barnabus Merrick, stayed some years at Leicester Hospital, and was subsequently employed by Sam Torr, the manager of a travelling show who exhibited all kinds of curiosities.

In 1886, Joseph Carey was taken to London Hospital where he became a patient of Dr Frederick Treves. The Elephant Man peacefully died on April 11, 1890 in the early afternoon. According to Dr Frederick Treves who made the postmortem examination, Joseph Carey died of a cervical
dislocation caused by the excessive weight of his head (Treves, 1885). Charles Barnabus, the only relative to have showed an interest to the boy, was in charge of the identification of his nephew’s body.

In 1981, David Lynch’s motion picture *Elephant Man*, with John Hurt, John Gielgud and Ann Bancroft as leading actors, won the Grand Prix of the Avoriaz Fantastic Film Festival.

**Suggested readings**

**Bibliography**

**Illustration**
Photograph of Joseph Carey Merrick in 1889. Taken from Howell and Ford, *op. cit.*