RESEARCH LETTER

Neurofibromatosis Type 1: Persisting Misidentification of the "Elephant Man" Disease

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Background: **During informal interviews in the course of an ethnographic study on intergenerational dialogue between individuals with neurofibromatosis and their parents, many members of Canadian neurofibromatosis associations have stated that they continue to be told the condition that afflicts them or their children is "elephant man's disease." Today, even though well-established clinical criteria make it possible to diagnose and differentiate the 2 diseases, the confusion between neurofibromatosis type 1 (NF1) and elephant man's disease persists in both the media's and physicians' representations.**

Methods: **This was an ethnographic study in medical anthropology.**

Discussion: **Some reference sources and print and online news media have all contributed to the persistence of the association between NF1 and elephant man's disease. Our observations suggest that confusing NF1 with the Elephant Man's condition harms the interests of those with NF1 and thus increases the burden of the disease.**

Conclusion: **Changes of attitude regarding medical teaching and the media could dispel the confusion among physicians and journalists. (J Am Board Fam Med 2011;24:112–114.)**

Keywords: **Elephant Man, Genetic Disease, Neurofibromatosis, Proteus Syndrome, Psychosocial Impact**

For many years it was thought Joseph Merrick, widely known as the Elephant Man, had suffered from neurofibromatosis type 1 (NF1).[1] In 1986, geneticists Tibbles and Cohen[2] demonstrated that Merrick was actually afflicted with Proteus syndrome, a much rarer condition. With an established diagnosis of Merrick's real disease and current knowledge about NF1, NFI should no longer be misguidedly identified with the disease the Elephant Man had. However, our recent experience in the context of an ongoing ethnographic study of intergenerational dialogue between adolescents with NF1 and their parents revealed that some physicians continue to identify NF1 as the same as Merrick's condition. This prompted us to investigate further, and we found that the confusion also persists in the current use of "Elephant Man's disease" as a synonym for and in media coverage of NF1.

NF1 is one of the world's most widespread genetic disorders. Its prevalence of 1 in every 3000 is almost identical to that of the far more widely familiar cystic fibrosis, with a prevalence of 1 in every 2500. In contrast, Proteus syndrome is a very rare condition, with a prevalence lower than 1 in every 1 million (Table 1).[3]

Confusing NF1 with the Elephant Man's condition harms the interests of those with NF1, all the more so because it is known that NF1 sufferers experience difficulty establishing social ties and developing good self-esteem.[4] Having their condition misidentified as the disease the Elephant Man had,

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Table 1. Comparison of Neurofibromatosis Type 1 and Proteus Syndrome

	Neurofibromatosis Type 1	Proteus Syndrome
Prevalence	Frequent (1 in 3000 to 1 in 4000 live births worldwide)	Extremely rare (<1 in 1,000,000 live births worldwide)
	Equally prevalent in men and women	Two men for every woman
Gene	NF1, chromosome 17	Unknown
Transmission	50% hereditary (autosomal dominant)	Sporadic
	50% sporadic (de novo mutation)	Postzygotic somatic mutation (embryonic lethal in nonmosaic form)
Diagnostic criteria		
General	Café-au-lait spots	Mosaic distribution of lesions
	Neurofibromas	Sporadic occurrence
	Axillary or inguinal freckling	Progressive course
	Optic gliomas	
	Lisch nodules	
	Characteristic osseous lesion (sphenoid dysplasia, thinning of long bone cortex with or without pseudoarthrosis)	
	First-degree relative with neurofibromatosis type 1	
Other		Cerebriform connective tissue nevus
		Linear epidermal nevus
		Asymmetric, disproportionate overgrowth
		Specific tumors before second decade
		Lipomas or focal atrophy of adipose tissue
		Capillary, venous, or lymphatic malformation
		Facial features including dolichocephaly, a long face, down-slanting palpebrae, ptosis, depressed nasal bridge, anteverted nares, and open-mouth position while at rest

and as a result being burdened with others' perceptions that they will grow profoundly disfigured over time, can only serve to increase these difficulties and compromise their hopes of achieving a normal social life, finding employment that interests them, enjoying an enduring romantic relationship, and having children.[1,5]

In 1995 the American anthropologist Joan Ablon[1] was the first to decry the continued association of NF1 with Elephant Man's disease by the media, yet the confusion persists. A striking example of media confusion relates to the first partial face transplant performed in France in 2007. The patient had NF1, but many French and foreign media stated he had the Elephant Man's condition. In the same manner, the term "Elephant Man's disease" continues to be treated as a synonym for NF1 in the entry at dictionary.com: Elephant Man's disease: "noun, neurofibromatosis".[6]

By expanding our inquiry, it would seem many medical professionals continue to confuse the Elephant Man's disease with NF1. On the one hand, some of the NF1 sufferers and family members in our study have told us that physicians continue to tell them the condition that afflicts them or their children is the Elephant Man's condition. On the other hand, when we make presentations about our study at scientific meetings, physicians approach us to tell us they are familiar with NF1 but they were taught that it is indeed the disease the Elephant Man had. That many physicians currently practicing were trained before the publication in 1986 of the Tibbles and Cohen[2] article may partly explain this.

Finally, some of the physicians we met with in the context of our ethnographic study held that this kind of confusion yields benefits in the form of publicity for NF1. But people who live with the disease believe the confusion reinforces prejudices about it and intensifies the sufferings of patients and their family members.

We believe that the medical and scientific communities that are responsible for the dissemination of knowledge should make a concerted effort to solve this problem. We must also find ways to better inform health and medical practitioners of

the clinical, ethical, and psychosocial problems that can result from confusing NF1 with the Elephant Man's disease. The factors that contribute to maintaining the confusion could be made known to those practitioners. Finally, a collaborative effort with the news media could dispel the confusion among journalists.

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