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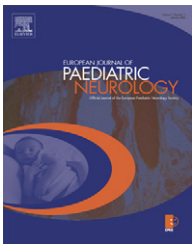

Original clinical historical contribution: Stuart Green Vignettes

Stuart Green's Vignettes 5 and 6

1. Some people don't tell

A young boy of 9 or 10 presented with a history of muscle weakness. He had been getting worse over a number of years. From the history it sounded as if it might be some form of hereditary muscle disease or peripheral neuropathy. I asked his father—a most articulate and rather Bohemian character—whether there was any family history and he said "No, not at all". When I examined the child he clearly had weak legs; they were thin with depressed reflexes, which suggested either some form of muscular dystrophy or disease of the anterior horn cells or peripheral neuropathy. The father then asked me whether I was "looking for a muscular dystrophy" as he thought this was a possibility. I asked why he had asked me this and he said that his own father had had weak legs, walked with crutches in his 40s and 50s. I then asked the gentleman himself whether he had any problems and he said "No, not really". I asked him if he had any difficulty in walking and he said "Well, some slight difficulty". I then asked him if I could see him walk with his trousers off. He was taken aback at this and said, "Why would you want to see that?" I replied, "Because it might be relevant". Very reluctantly he removed his trousers and there as clearly as could possibly be seen was a man with thin legs who walked a slightly awkward way and yet he had clearly said that there was no family history. He was intelligent, a rather unusual man and I asked him why. He had some odd idea that he had picked up an infection in the Army and had transmitted this to his son. He thought if he hadn't told me anything about the family at all and didn't mention his past history, I wouldn't have diagnosed anything.

Comment

Stuart extracting a family history reminds us that elicit may mean to squeeze out. There seem to be several reasons why information on hereditary disorders is withheld, and eliciting a complete family history is one of the most difficult tasks we have. This vignette illustrates that every possible family member may need to be not only interviewed but also personally examined.

2. The PKU boy who didn't respond

A 5-year-old boy with phenylketonuria diagnosed at birth had been poorly managed by diet because his family "couldn't cope". His mother had separated from his father, his mother was said to be "odd and incapable" and the details of his father were not known. He was adopted by an elderly couple and was put back on to the diet. His diet was well monitored and he really did very well. He thrived. At the age of 5 he was almost back to normal with good gait, good coordination and reasonable speech. However, over the next year he started deteriorating and became in some ways uncoordinated in his speech and became clumsy.

He was investigated extensively for all forms of variant forms of PKU but none of these proved positive. A very experienced PKU nurse said she had worked with a lot of PKU cases where the child had been mismanaged on the diet and this boy looked nothing like these at all.

MRI imaging showed two sets of abnormalities. One was fairly typical lesions on the cerebral cortex as in PKU; the other lesion was in the basal ganglia (deep parts of the brain concerned with motor control).

We did very extensive screening for rare causes of incoordination and to our great surprise and sadness he turned out to be positive for Huntington's chorea. It turned out that his mother who had given him up for adoption was not incompetent because she had Huntington's; the mother couldn't cope with the family situation because the boy's father had had Huntington's! He drank excessively and she found the whole situation too stressful. Thus, this boy had the very unfortunate combination of two rare diseases causing much devastation for his adoptive parents. They however coped with the news remarkably well and 2 years down the line fortunately he is still stable although uncoordinated and his speech has deteriorated.

Comment

Paediatric neurologists are always glad to find a rare—or not so rare—treatable disorder. When an unexpected coincidence occurs it is beguilingly easy to succumb to Kouska's fallacy,[1] a trap into which Stuart would not fall. Rather, this vignette illustrates Stuart—a clinical doctor—unravelling the complexities of family life.

As commentators on these vignettes we have not altered Stuart's wording, but sometimes we add a clarification that feel Stuart would have approved of. In this case the typical lesions of PKU are in the white matter and are more complex than we used to think.[2]

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