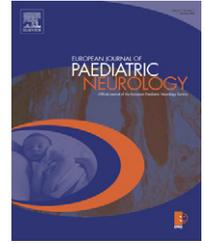


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## Original clinical historical contribution: Stuart Green Vignettes

### Stuart Green's Vignettes 3 and 4

#### 1. You have got to see it

One of the problems of paediatric neurologists dealing with epilepsy and other paroxysmal (occasional) events is that one is entirely dependent on the observations of the parents or the child themselves about exactly what has happened. After all a surgeon dealing with breast lumps or hernias examines people and “sees” what is there but many children we see in paediatric neurology are on examination entirely normal and it is only the character of the event that gives the diagnosis. This was the position until recently when people started to video events, and this has made an enormous difference to this area of paediatric neurology (see later).

A young boy of 11 or 12 years presented with a history that he would suddenly stiffen particularly when he moved suddenly. These episodes would last a minute or so and then he was all right. It almost seemed, from the way he described it, as a sort of cramp but he was not able to describe it very well and his mother was rather reluctant to imitate his abnormal movement. (It is surprising how many people are unwilling to actually reproduce or imitate the abnormal movements of a child. They will try to describe them but they won't imitate. Perhaps they are embarrassed.) In any event the boy was entirely normal, we were not able to reproduce the movements. However as the boy got out of the chair to go he suddenly twisted his neck, stiffened his left arm and turned his body round in a grotesque posture which lasted 20 or 30s and then it stopped. I immediately recognised the condition, a so-called paroxysmal dystonic movement, one called paroxysmal kinesigenic dystonia in which in some way the initiation of movement comes about in abnormality of posture. Recently, it has been thought that this condition and a number of other similar conditions are due to so-called channelopathies, that is conditions in which the ingress and egress of sodium potassium from the cells are defective.

I asked the young man how often he had this problem and he said he had it two or three times a day. I asked if it was a problem for him and he said it was not much of a problem but was embarrassing sometimes. Then his mother interjected and said of course we would like him treated because it must be quite painful for him. I then asked her why she thought it was painful if he himself had not said

so. She said to me—and the conversation went something like this—

Mother—“It is obvious that it must be painful to be in such a position”

Dr. Green—“But he didn't say so himself”

Mother—“But I know it must be”

Dr. Green—“How do you know that?”

Mother—“Because when I had it, it was very painful”

I stopped in my tracks. “When you had it” I said to mother. “Yes- I had it from the age of 10 to about 23. It stopped when I had my children”

Dr. Green—“What did the doctor think it was”

Mother—“I don't think I ever saw a doctor”

Dr. Green—“What did your parents think about it?”

Mother—“Well it didn't happen very often and I used to try and hide it with other movements. I never actually told my parents, so nobody knew about it”

Dr. Green—“So you are telling me you had this abnormal movement for 10–15 years and now its gone away, but you never mentioned it to me until just now”

Mother—“Well I never really thought about it until we were talking about the subject and I really saw what happened to my son”

I am pleased to say the son was put on to carbamazepine and he did very well. Although it is standard teaching always to take a family history, patients and relatives do not always tell you (for a variety of reason) the critical information which gives a diagnosis.

#### Comment

Listen and imagine because it is almost never there to see during the consultation! (and even nowadays the parents may not have videotaped the whole event or its most important part: the onset and the end). The diagnosis of paroxysmal neurological disorders which are so numerous (besides epilepsy) is probably our most important clinical activity and nothing will replace the description of the event and the art of bringing it to life again by history. We are sure that Stuart Green would have made the diagnosis, even if the boy had not had an attack then and there. Or did he know he would have one and was prepared to see it?

But would the mother have told about herself without him asking the boy how he felt during the attack?

## 2. The internet revolution

Over the last few years the spread of the internet has had a profound effect on medical practice particularly in the field of paediatric neurology. For many children who have rare diseases parents have more time to look these up and find out more about them than the poor overworked doctor does. These few stories indicate the complexity of the problem. A man whose child I had been following up with cerebral palsy came in to me almost in tears and said “Dr. Green, my wife hasn’t slept for four weeks since you last spoke to us about the diagnosis.” I asked why this was and he replied “Well, we are terrified because she is likely to die of heart failure from Friedreich’s ataxia.” I was surprised—looked at my notes and said “I’ve never mentioned Friedreich’s ataxia (a progressive disease causing stumbling, falling, weakness and heart disease). He said “Well we looked it up on the internet and it said that these people often die in late teenage or early adult life with heart problems”. I said “That’s true but I never said your child had Friedreich’s ataxia”. He said to me “You said she had ataxia” I said “Well, there are literally hundreds of different types of ataxia. I did say that your child had cerebral palsy (a non-progressive form) of incoordination probably arising before birth and nothing whatsoever to do with Friedreich’s ataxia.” He said “Well we found it on the internet under ataxia and you never told us it wasn’t. You should have told us it was *not* Friedreich’s ataxia!”

A rather more sophisticated man whose child had a generally complex problem of a progressive dystonia causing abnormal posturing and twisting of the limbs which can be due to a wide variety of genetic and acquired conditions, came to see me for a further opinion. His opening words were literally “I’ve seen my local consultant and I’ve been down to Great Ormond Street I have done a great deal of reading on the subject. As far as I understand there are 39 separately identifiable causes of dystonia, but only 32 of them have been excluded I would be grateful if you could exclude the other 7 (I didn’t have the heart to tell him I didn’t think I knew more than 20) so I said to him that I would try my best.

A lady presented to me with a youngster having transferred from another town, with unfortunately severe retardation due to a condition called lissencephaly in which the brain is abnormally underdeveloped and smooth. It is a rare condition

although there has been recent research which has increased our knowledge of it. She seemed satisfied with the consultation and I was impressed with her knowledge of the subject. I said to her that there was a fair amount of new information on this condition and there was a support group. The mother said “Yes, I know that Dr. Green” and I said “Have you seen the literature yet? I think it’s very good” and she said “Not bad”. I said that there was a particularly good review in the last Newsletter, and she said “Yes, I know”. I said to her “Are you a member of the group?” and she said “Yes”. I asked here if she was on the Council and she replied that she was. “Did you write the last Newsletter?” I asked and she replied “Yes”. I asked if she had written the article and she said “Yes”. I had met my match!

### Comment

When the internet became widely available and routinely looked at for all kinds of information including medical ones, many of us thought that our clinical life and communication with parents would become impossible for the reasons illustrated here. Since this piece was written probably 10 years ago, we tend to ask parents very soon if they have already looked at the internet and what they have concluded. More often, a feeling of confusion, anxiety, the sense of the complexity of the medical problem and the contradictions between the uncontrolled multisources they found is the response. On the whole, this has not made our life more difficult as we had anticipated, but often in fact easier and our work more respected. Not everybody will agree!

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