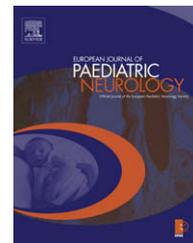




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Original clinical historical contribution: Stuart Green Vignettes

Stuart Green's Vignettes 13 and 14

Thierry Deonna^a, John B.P. Stephenson^{b,*}

^aDepartement Medicochirurgical de Pediatrie, Unite de Neuropediatrie 1011, CHUV, Lausanne, Switzerland

^bFraser of Allander Neurosciences Unit, Royal Hospital for Sick Children, Yorkhill, Glasgow, Scotland G3 8SJ, UK

1. Doctor not up to expectations

A boy of 6 years was diagnosed upon an acute collapse as having insufficiency of the adrenal glands, which was subsequently diagnosed as part of a very serious degenerative disorder called adrenoleucodystrophy, in which the children (boys only) develop a combination of adrenal insufficiency (which can be corrected using hormonal treatment) and progressive loss of intellectual and physical skills which at the time of writing is still difficult to treat. They very often, but not always, die early in life. This condition made famous by the film *Lorenzo's Oil*, which is a possible palliative treatment. A more recent possible treatment is a bone marrow transplant. The bone marrow donor must have similar tissue factors to the child (like blood groups) and of course must not have the gene for the condition itself.

The family were clearly aware of the genetics of the condition (that is manifested by boy as and passed through the mother) and the factors associated with bone marrow transplantation and the risks involved. They had understandably been very anxious about this condition and whether or not the younger boy – now aged 5 years – should be tested. One important point about this condition is that not all children who have the genes for this condition necessarily manifest the disorder – only about 50% do.

The difficulty of assessing any particular therapy – at the time of writing it was thought appropriate to transplant people who had progressive symptoms (which the younger boy did not yet have) and inappropriate to transplant them if they were normal (which he was at the moment) but probably appropriate in a small window when they began to deteriorate. There was some suggestion that he might be deteriorating and the question of marrow transplantation came high on the agenda.

They had after some discussion expressly declined that the younger brother should be tested so we did not know whether he carried the potential for the disease or not. I then had a long discussion with the father about the possibility of transplantation and he asked me why it was not possible for the younger boy (whose status we did not know) to be used as a transplant donor. I said that we couldn't use him as a transplant donor because we didn't know whether he had the condition or not (both the parents had expressly forbidden us to test him for it.) The father, who was quite a senior civil servant; was extraordinarily dismissive of my comment and said that he was extremely surprised that a Unit like our which was meant to be a tertiary referral centre, did not know after all this time whether or not his younger son was likely to be affected or not. He paid scant attention to my limited protestations that he himself had refused for his son to be tested. In my experience, however intelligent people are, when they are very anxious they are very rarely logical.

1.1. Comment

This is the kind of difficulty that all clinical paediatric neurologists experience. It illustrates why we need so much time with our patients' families, though even with the best care matters may not turn out as we would have wished.

2. The symptoms give the pathology

Sometimes in neurology – maybe it happens more often than we realise – we learn about the way the organisation of the nervous system is structured by the way it breaks down.

A boy of about ten years was travelling with his family in a car and said he had a bad headache. His mother gave him

* Corresponding author. Tel.: +44 141 7765589.

E-mail addresses: thierry.deonna@chuv.ch (T. Deonna), john@jbstephenson.com (J.B.P. Stephenson).
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a drink of water and told him to lie down in the back of the car. A few minutes later he said he had a very bad headache and his father said they would stop at the next service station to get some paracetamol or analgesic.

A moment or two later he said I have such a bad headache I think I am going to die. His parents were naturally petrified at this and they rushed him to the nearest hospital at which time he had collapsed. A CT brain scan showed he had had a bleed into his occipital lobe and he was transferred urgently to our Neurosurgical unit where the neurosurgeons operated and took out quite a large haemorrhage which they believe was due to bleeding from an arterial venous malformation (a malformation of the blood vessels which had spontaneously bled). He had no other abnormal bleeding vessel and he slowly and gradually made a recovery.

About three or four days he was out of intensive care and by two weeks he was on the ward and was walking with somewhat of a limp. He could understand people but he couldn't speak very well. His behaviour was rather erratic. About a week or two later his speech had improved considerably but he still had difficulty in walking on one side; he couldn't see to one side of his visual field but the major problem for the nurses was his hypersexual behaviour. He was exposing himself, he was touching nurses and his parents became very distressed about this. This became the major problem in his management.

The damage to his occipital lobe had caused him to have a visual field defect. This is relatively simple hard-wired but because the visual cortex was destroyed in this area and it is

so specialised the other side of the brain could not take over and he was left with a permanent visual field defect towards to right hand side. The haemorrhage had spilled over and caused some damage to his parietal cortex which controls language. There was some recovery from this area, partially because that part of the brain was incompletely damaged and probably partially because although at the age of 9 there was some possibility of recovery the other side of the brain may have been able to take over certain functions. His hypersexual behaviour was more difficult to explain. It was partially to do with his age, his sex, his state of development, his past history, the fact that other areas of the brain had been disturbed but not initially damaged by the event and gradually over a number of years this settled down and his behaviour became more normal. He was left in end with a dense hemianopia, a limp, minimal language problems and tolerable behaviour.

Although the brain is remarkably adaptable there are certain key areas which if damaged cannot function. In young people one side of the brain can take over language function to some extent, usually more so before the age of 7, prediction of what will happen to children from brain damage is notoriously difficult.

2.1. Comment

No further comment is needed after this elegant description by Stuart of one of the child patients who like us he saw every day.