

Great Ormond Street Hospital for Children NHS Trust

and the Institute of Child Health

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Medical Report on

Bethany MILLS dob: 20.09.96

6 Parry Close Glendale New South Wales 2285 AUSTRALIA

Bethany was seen for consultation in Out Patients on 4th August 1999.

She is a two year old girl. Pregnancy was normal. She was born at thirty-nine weeks by elective Caesarian section because of cephalo-pelvic disproportion. There were no immediate neonatal worries and she developed mild jaundice which needed no treatment. Her early cognitive and developmental milestones were normal.

In August 1998 she developed a limp in the right leg which improved spontaneously. In February 1999 at the age of two and a half years she developed a fever and some diarrhoea and vomiting. Overnight she developed dystonic posturing of the right-hand side that over the next few days also involved the left-hand side. Recently there has been some improvement in this and abnormal posturing is only noted on the right. There was no apparent improvement in the dystonia with Co-careldopa which caused significant nausea and vomiting. Her development continues, although she is said to be slightly backward in pre-school.

Apart from her sister, there is no family history of dystonia and Parkinsonism. Her father has attacks of an odd feeling in the throat associated with an inability to breathe that lasts for approximately one minute and occurs approximately monthly. Her older sister, Leanna, also has dystonia and an older sister, Katie, has an attention deficit hyperactivity disorder. Bethany currently takes

Examination: Head circumference was normal at 46.5cm She is not dysmorphic and her skin was normal. Both saccadic and smooth pursuit eye movements were normal. Her ocular fundi were normal. The rest of the cranial nerves were normal. In the limbs, she had a mild right hemi-dystonia that increased with intention. Her deep tendon reflexes were all brisk, she had toe jerks and her plantar response was up-going

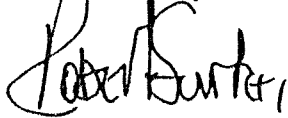
Opinion: I think that Bethany has a primary dystonia. The asymmetrical onset, progression over months and the lack of orobulbar involvement are all typical of idiopathic torsion dystonia of childhood. Approximately half of children with idiopathic torsion dystonia will have the common mutation in the DYT-1 gene and this is inherited as an autosomal dominant with highly variable penetrance. Children with DYT-1 negative idiopathic torsion dystonia may have inherited this in either a dominant or a recessive manner. The results of testing for the common mutation in the DYT-1 gene are still awaited. Bethany has been extensively investigated in Sydney for all the known causes of secondary dystonia and these investigations have been negative.

I would recommend that the following drugs could be tried

1. Benzhexol, starting in a dose of 0.5mg three times a day and increasing by 1mg a day every week until one of three things happens: (a) she gets better; (b) she develops side-effects (commonly dry eyes and mouth or constipation); or (c) a total daily dose of 40mg is reached.
2. Tetrabenazine, starting in a dose of 12.5mg three times a day and increasing at two to three-weekly intervals to 25mg three times a day and then 50mg three times a day.
3. Pyridoxine, 50mg twice daily
4. Phenytoin in its usual antiepileptic dosage
5. Solperide
6. A combination of Solperide and Tetrabenazine.

For these trials, each drug should be given in the highest tolerated dose for at least three months before concluding that it has been ineffective and the next one tried.

Yours sincerely



Robert Surtees

Senior Lecturer in Paediatric Neurology