How bad can it get?

We are writing as parents of a young girl who “suffers” from precocious puberty and we use the word suffers without reservation despite the fact that:

• Puberty is a normal process
• No underlying cause triggering her puberty has been determined.

Our daughter started to show physical signs of development, for example, breast development combined with pubic hair, at 6½ years, and within a year experienced her first period. We were devastated by this event, as our local general practitioner had told us that there was nothing to worry about, and that we were imagining and exaggerating things. Our ongoing concerns for an explanation and regular visits to the surgery were seen as pushy and demanding. Surely confronted with this period, an obvious sign of female maturity arriving very early, we would get help and support. Regrettably, it didn’t get any easier.

The good news was that the general paediatrician to whom we were first referred was really helpful. He confirmed that he had rarely encountered the condition before but would organise particular tests to discount any underlying causes for this early growth, such as cranial or ovarian tumours. Our shock with these possibilities was diminished by the speed of his tests and his effectiveness in communicating to the three of us as results came in. Fortunately no underlying cause was identified and we were referred to a paediatric endocrinologist. Confusion was about to reign again.

The specialist recommended that our daughter, who has “central precocious puberty”, should receive analogue treatment (with gonadotrophin releasing hormone analogue) to suppress her accelerated development and enhance her height potential. Growing up early, he explains, will result in compromised final height. Before commencing this treatment we seek a second opinion from another paediatric endocrinologist. We find that an insufficient range of tests have been organised to discount any underlying cause. We need to reaffirm the diagnosis before starting treatment with such an analogue. The treatment may now be irrelevant, but in any case it doesn’t, in his view, enhance final height attainment.

“Now the shock is turning to dismay, anger, and frustration”

Eighteen months of consultations and divided opinions pass before we can commence treatment—during which our daughter is continuing to develop. Monthly periods, moodiness, greasy skin, acne, but most critically accelerated bone maturation, reducing the scope for further height attainment. At last, our third specialist consultant agrees to organise the treatment and we proceed down this new route.

Our daughter’s periods stop, her skin improves, moodiness reduces, and our child comes back to us. She even prefers an injection to a period and her confidence with her peers returns to previous levels. She is normal again! But to what end and at what cost? Our daughter finished her 12 months treatment early this year and her recent final bone age test now confirms that “too little was done too late”. So what is the net effect after three years of consultations and divided opinions pass before we can confirm the diagnosis before starting treatment with such an analogue. The treatment...
development is becoming more common in recent years, but the evidence for this is poor, as shown by Viner.1

Most general paediatricians confronted with a young child showing signs of puberty would initiate early investigations, such as checking gonadotrophin and oestriadiol levels, thyroid function, β HCG, prolactin, and a bone age and a pelvic ultrasound. Depending on local facilities they may well organise a magnetic resonance imaging (MRI) scan of the mid brain and pituitary area. They would determine whether the sequence of puberty development was normal, plot growth carefully, exclude any signs of raised intracranial pressure and look for any abnormal pigmentation to suggest McCune–Albright syndrome. It is likely that a second opinion will be sought from a paediatric endocrinologist. One of the key things during this difficult time for the parents is effective communication of what is going on, and what investigations are to be undertaken and why.

The paediatric endocrinologist should be in a position to continue the discussions initiated by the general paediatrician, decide whether further tests are needed, and will no doubt discuss the issue of blocking treatment. Unfortunately, as shown at a national meeting on premature sexual maturation organised by the Child Growth Foundation in London in 2000, there was no strong consensus view among endocrinologists about how extensive investigations should be and how aggressive analogue blocking treatment should be with gonadotrophin releasing hormone analogues. Some endocrinologists argue that MRI brain and pituitary scanning has a very low pick up rate in girls unless very young, and there was disagreement about whether this test should be undertaken at 7–8 years, although nearly all agree that at 6½ years girls should have an MRI scan, as well as all boys below 9 years.

Views about whether children should have blocking treatment were more variable, with quite a wide range of opinions; it was pointed out that there is no strong evidence base on which to judge these decisions. The issue of which patients to treat with analogue treatment is discussed by Brook2 in his textbook on paediatric endocrinology; he favours treatment in younger patients. His view is that the question of treatment or no treatment should be based on assessment of growth prognosis and the child’s psychology maturity. The psychological aspects of the condition on a young child and their family are easy to underestimate by professionals.

If treatment is to be given, there is now another controversial issue of whether a standard intramuscular injection on a 3–4 week basis should be given, or whether a slow release preparation should be given every 2–3 months—the efficacy of the latter was shown by Patterson and colleagues.3 In the past the issue of advanced bone age was given as a strong reason for starting treatment to prevent premature fusion of the epiphyses and short final height stature, but the evidence for this in children with later onset is inconclusive. The parents at this meeting felt it frustrating and difficult to accept that doctors from different teaching hospitals had divergent views. With this background the issue of communicating well with parents is crucial.

In the case of this young girl the parents mentioned that a second paediatric endocrinology opinion was sought. It is not clear why they felt this was necessary, but it did result in delay of treatment which was eventually commenced 18 months after onset of periods. The parents state candidly that it doesn’t take much to imagine their views of the medical profession. The Child Growth Foundation has a difficult role as it needs to support parents with this frightening condition, give background literature and information, and sometimes give suggestions of where to go for second opinions. There is of course an inherent danger in second or third opinions, as there may be conflicting views which may cause confusion and delay treatment.

The survey carried out by the Child Growth Foundation is helpful:

- General practitioners should certainly refer urgently and listen to parents.
- As discussed, the roles of paediatricians and paediatric endocrinologists need to be distinguished.
- Should analogue treatment be given? Further research is needed to look into the issue of whether it enhances growth potential. Many would agree that the growth argument with premature fusion of the epiphyses has been unintentionally exaggerated in the past.
- The parents ask about help for the child and the parents in dealing with this difficult situation at school and socially; this must be a key role for the Child Growth Foundation. Hopefully most paediatricians would look in the CaF (Contact a Family) directory to look up the support groups in such a situation. Community paediatric nursing teams also have a support role in giving advice at school and at home.
- How can the professionals assist with giving proactive advice and assistance?

Have a look at the full survey by the Child Growth Foundation and their Best Practice Guidelines. Treatment for this rare but devastating condition for parents and children is still somewhat unpredictable. Do write in with your views and comments.

N Mann, Commissioning Editor

REFERENCES