Living with genetic rickets

Unless you actually have a metabolic bone disorder like hypophosphataemic rickets, you don’t really know how it feels. Even if every member of your family has this syndrome, and so you experience the effects by proxy, you still can’t feel it “in your bones”. But if you’re the odd one out trying to understand how your family members feel, you can still be an important help-mate, and that’s what this article is about. I want to describe how, within my family I have been coping with the chronic condition that affects us all.

The treatment for this particular kind of vitamin D resistant rickets (actually a historical misnomer, because many people affected by what is now called X linked hypophosphataemic rickets do, in fact, respond well to the active metabolite calcitriol, though processing of the native form, calciferol, is poorly regulated; another form of genetic rickets, involving the vitamin D receptor, is truly D resistant) has been pretty consistent over the past two decades. A combination of oral phosphate and capsules of calcitriol form the approach that seeks to balance or redress the levels of inorganic phosphorus, calcium, parathyroid hormone, and alkaline phosphatase. Because this management regimen often manages to achieve near normal blood chemistries, the epiphyseal fray of patients’ bones associated with rickets often heals and over time their bowed or twisted lower legs may straighten as well.

Indeed, this was the progression we proud parents observed after several years of our daughter’s growth. From being severely bowed at the age of 3 in 1986, with a pronounced penguin waddling gait, she became so normal looking by 1989 that finally she was deemed unsuitable for scrutinising candidates for the Paediatric Boards! If that were the end of the story, however, I would not be writing this now.

Was it a combination of pre-pubescent weight gain, or heavy school satchels worn over one shoulder that conspired to foment a increasing wind-swept tilt in our lovely daughter’s legs as she grew towards adult height? Whatever happened, from being complacent about the resolution of the disorder, we were suddenly challenged in 1994 when we realised she couldn’t run anymore that our daughter’s legs had become severely compromised, so that corrective surgery was the only option ahead.

Our son, born in 1987, and identified with the syndrome early on, had never quite reached above the 3rd centile for height, even though his managed chemistries were always satisfactory. But being short, even very short, is a situation you can cope with, as long as your legs are straight and you can play sports. Sadly, as he grew older, his lower legs did not keep pace, and progressive bowing and in-toeing meant that his participation on the team playing field was never going to be felicitous. Meanwhile, he could experience by proxy as much as a 10 year old is able, some of the implications of his sister’s surgery.

Being of practical and optimistic-looking disposition, we anticipated the surgery with hope and some dread, as her mother and aunt had both dealt with their tibial osteotomies with courage and good resolution. It was around this time, some seven years ago, that as the only unaffected family member, I resolved to learn about the syndrome as best I could. Learning was one thing, and since I was an academic-associate at university, I placed my findings on my own website that I created there. The site proved to be a magnet for finding, like needles in the haystack, other families affected by this rare disorder, and within several months a patient support nucleus was formed.

This support network was invaluable in keeping me reasonably calm and focussed during our daughter’s surgery and recuperation. Since the major bend was in her femur, her whole leg was immobilised, and she was off school for an entire term. As frantic as I was, I found that the support network was a place of solace, with other families pouring out experiential information, empathy, and consolation. I found comfort too (in being useful) with a home schooling network, and the use of mind-mapping as a study guide proved valuable for our daughter’s intellectual development as her leg recovered.

Since it was just around the time of inception of this XLH Network that the gene (PHEX) affected in XLH was identified (no doubt our family was an important cog in helping to isolate the specific locus), there was additional incentive to learn all about the basic biology of the disorder. Everyone knew that treatment could be useful, but that in some cases it was less than satisfactory. So it was eagerly anticipated that gene identification would provide quick answers for a real biological therapy.

Seven years down the road, the whole story of phosphate homeostasis and genetic rickets is now intensively analysed (usually as a means for biotechnology exploitation and pharmaceutical intervention in the more common osteoporosis syndromes), but concrete understanding is still in a state of flux as academic researchers wrestle with fiendishly complicated positive and negative feedback loops. Everyone hopes that a five year horizon is realistic in terms of finally understanding why bones don’t grow well when you have XLH, and identifying the very best approach to treatment.

The XLH Network (now professionally present at www.xlhnetwork.org) is growing too, as it encompasses nearly 400 members around the world today, and we as an affected family feel empowered that we are doing as much as we can to help to understand and manage this disorder. Because of the network, we helped to bring phosphorus tablets into the UK, which may enhance compliance by avoiding the noxious slimy taste associated with the effervescent phosphate solution of the past. Because of the network, we have helped to publicise the commercially available
genetic test for XLH, while considering just what this sort of genetic testing can mean to affected families. Because of the network, we are much more confident in our children’s treatment because it is obviously the approach of the best centres. Because of the network, we know more about osteotomies, Ilizarov apparatus, bone pinning, stapling, and cajoling, so that while the trepidation is still there, we face our son’s imminent tibial osteotomy with reasonable confidence too.

Managing a chronic condition successfully may involve every member of the family, since even the ones who aren’t directly affected still feel deeply for their loved ones, and for their own self-esteem and continued good health, they do need to be involved too.

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IMAGES IN PAEDIATRICS

“Fixed intestinal loop” on abdominal x ray: sine qua non of mechanical intestinal obstruction

A term neonate underwent primary closure of gastroschisis and limited small bowel resection for a perforation, in another institution. Four weeks later, gastric aspirates remained high and he developed abdominal distension. Contrast enema did not demonstrate any obstructive lesions. Plain abdominal x ray examinations on two subsequent days demonstrated a fixed, dilated intestinal loop (fig 1). At laparotomy, he was found to have extensive adhesions and complete occlusion of his previous anastomosis, with a dilated and atonic proximal segment. A limited resection with primary anastomosis was performed.

Persistent “ileus” obstruction following gastroschisis repair and a delay in commencing feeds for 20–30 days is common in gastroschisis babies.1 The key is to distinguish between mechanical and functional subacute intestinal obstruction, as management will differ.

Radiological investigations play a vital role in the management of intestinal obstruction in all ages. Radiologically, the mid-small bowel is the most inaccessible and nondescript part of the gastrointestinal tract. Both mechanical and functional obstruction of the mid-small bowel can appear as non-specific dilatation on plain abdominal x ray. A fixed and dilated loop of bowel that does not alter in size, shape, and position on repeated abdominal films is highly suggestive of mechanical bowel obstruction due to an aperistaltic segment of bowel (usually due to ischaemia or gangrene). A typical example is in necrotising enterocolitis which mainly affects premature babies. It is a strong indication for surgical intervention and should not be overlooked.

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Reference