

From NZAN newsletter No.25, December 05

## **Joel's and Joshua's Story Diagnosis and stabilization**

Seventeen year old Joel was at the [December 05] Auckland meeting with his twin brother Joshua, and his mum Sandra.

Joel's diagnosis of Addison's disease was confirmed in July. His mum took him to the GP in April, concerned about a lot of symptoms developing since Christmas - weight loss, fatigue, sleep problems, thirst, feeling cold, irritability, inability to handle relatively minor infections. He'd been very sick with the flu, and also after the meningococcus vaccination. Sandra was wondering why Joel's eczema scar tissue especially inner thigh and behind his knees was turning very dark. [He did have some salt craving, which she recognised in hindsight.]

Joel's GP promptly referred him to the general medical team at the Counties Manukau Clinic at Middlemore Hospital. "While we waited for an appointment, I took him privately to see a paediatrician. Unfortunately he didn't pursue Addison's, even though [I found out later] Joel's serum sodium result was low, and a 9am serum cortisol was in the low range, generating the comment on the form to consider a short synacthen test for adrenal insufficiency.

The paediatrician's letter to Joel's GP commented on his good colour, and included 'I cannot find any distressing abnormality with Joel, and hope he still continues to grow to achieve his height potential.'

Fortunately, the appointment for the hospital clinic came at the end of June. Their GP encouraged them to attend for a second opinion. At the Tuesday clinic, Joel was seen by a locum, whose uncle has Addison's disease. Joel was sent for an ACTH stimulation test, diagnosed, and started his meds on Friday night.

He was referred on to the endocrinology clinic, and seen there. Then, the family decided for accessibility reasons, to transfer to an endocrinologist whom they see privately, who has monitored both boys "He has been very accessible with phone advice, scripts, and prompt appointments when needed, which has made the adjustment much easier for the whole family to deal with."

Although he was symptom free at the time, twin Joshua's ACTH stimulation test showed some cortisol reserves, but a blunted response. Like his brother he had positive adrenal autoantibodies. It was considered just a matter of time.

Mum kept a close watch. First, Joel's symptoms had shown in contrast to Joshua's continued good health. Now, with the comparison of Joel stabilized on

his meds and well again, Joshua was showing the same symptoms that had led to Joel's diagnosis.

A few days after the NZAN Auckland meeting in December, Joshua awoke feeling hot and unwell. He'd been feeling wrung out and exhausted all week. He didn't want to get out of bed to go with his brother to help Dad at work - something he enjoyed. Mum realized it was time for some hydrocortisone, same morning dose as Joel, as the specialist had primed her. He was still not wanting to get out of bed an hour or two later, and so his mum gave him a repeat 15mg of hydrocortisone. A trip to the GP confirmed the presence of an upper respiratory tract infection, and he started antibiotics.

Sandra emailed Jeanette later that evening:

"He is soooo much better already. I gave him another 20mg late afternoon and he is currently sleeping like a baby after a comparatively busy evening talking to all his friends on the phone - that's more like my boy!!.

"Today really showed me the impact that low cortisol has on Joshua's behaviour and sense of well being. Addison's sure does present rather a big learning curve! I think Josh understands things a bit better too after today. He commented this afternoon that **having low cortisol was like having a well trained and equipped army but without any ammunition to fight with.** I thought that wasn't a bad metaphor for one so new to this game!"

A few days later Joshua was checked over by his specialist, who'd fortunately had a cancellation. All things considered, he agreed it was time for Joshua to take daily replacement meds, the same hydrocortisone as Joel [15/5/5], but 0.1mg fludrocortisone rather than 0.2mg for now.

Sandra tells us that their GP is a very good listener, and certainly willing to learn about Addison's. " I am very grateful that he referred Joel on as soon as he presented with symptoms. He has been wonderful moral support since diagnosis.

Sandra is also pleased to have talked with another mum whose twin boys were diagnosed some months apart 14 years ago - when aged 11.

