MR SMILEY ☺

"I'm sorry, Mrs Slender, but the results of your son's tests have come back and he has Trichothiodystrophy! I'll send you some information in the mail and the Geneticist will be in touch. Have a good Christmas."

We received the two pages of information about this disorder that none of us had ever heard of and began the search for information.

After that Christmas, I took James to visit the Geneticist, and I asked my questions.

"Is the delay only physical?" "No."
"Will he walk?" "Maybe."
"Will he have an intellectual disability?" "Probably."
"Are his skin problems part of it?" "Maybe."
"Are there any medical issues?" "Definitely."

Questions and vague answers continued for some time before I asked if there was anything else she could tell me and her answer, which still goes around in my mind to this day, was "Don't expect too much."

That statement began a 16 year journey of ups and downs, but we were determined our son would reach whatever his destiny was. Today, we still don't know a great deal about Tricothiodystrophy. It's one of those rare disorders that doesn't attract funding for research because it affects so few children.

Our association with Mater Dei started when James was two years old and we were making weekly visits to its Bethany Early Intervention Service for therapy, advice and friendship. Unfortunately, due to my work commitments, James had to leave 'Bethany' and continued his therapies under the watchful eye of our local occasional care centre and preschool.

All the while James was developing slowly. Some milestones were being met, and these were exciting moments. We could say "Yes" to some of the questions that we were being asked, at last! "Yes, James does talk, in short sentences… yes, we can understand him" and "Yes, we can have a conversation with him." Now we say, "James, please be quiet!"

Looking back, there are many memories from his primary school years. From the time James wouldn't talk to me for a long time after major surgery on his legs, because his legs were so sore… to the time when he ran down the track at Homebush Stadium (when he raced for his local centre) with the biggest grin on his face and shouting out "I did it, Mummy, I did it" , to the time when he played soccer for the first time "Just like my brother", to the time when we watched him address the Support Unit Presentation Day Assembly when he was in Year 6.

James was growing up and now ready for high school. What do you do? Where do you send the special little person you have protected for so long? It was at that time we crossed paths with some friends we hadn't seen for a long time, Garry and Kathleen Saunders, whose son Brenton attends Mater Dei. To cut a long story short, we ended up back at Mater Dei.

When I look at James today I really wish that Geneticist could see all that our son has achieved. From that very moment of diagnosis, we have never stopped looking for treatments, therapies and educational settings which have all contributed to helping James reach his potential and become the amazing person that he is.

Today, James has grown to become a confident, happy and independent young man. At school, he has embraced the many opportunities that have been offered to him and particularly loves being a member of the Mater Dei Choir, featuring in any performing arts events, helping out in the Mater Dei Meals on Wheels team and being a resident in the Living Skills Program, two nights per week.

Sometimes the decisions have been difficult and ones that we would have preferred not to have to made. Having a child with special needs is scary, heartbreaking, demanding and rewarding… James' cheeky smile, his constant chatter and the always-busy little person will be with us forever. Our home is always filled with the joy and innocence of childhood and the constant discoveries that a child makes.

Sharon Slender