

# Charlotte Redwood

## Age 9 years Old

### Case History

As Charlotte's mum I did not experience any complications during pregnancy or birth. I went into labour two weeks before due date and was given an elective C-Section due to difficulties experienced with my first born. Charlotte was breast fed and had no obvious feeding difficulties.

Just before her 2nd birthday we had a report from Charlotte's nursery that she was behind her peers in some areas. After our health visitor did a quick review she referred us to Speech & Language services who then transferred us to the Boost Centre at the RUH in Bath. The group of professionals (OT, Physio and SALT) could see delays so gave her weekly sessions for blocks of six weeks term time only. This continued until she was 3 and a half years old. From that time Charlotte had a diagnosis of Global Developmental Delay. Her speech, language and communication skills were very severely affected.

At 7 years old our consultant enrolled Charlotte on to the 100,000 Genomes Project where we later received a confirmed diagnosis of what has caused Charlotte's difficulties. Charlotte has a very rare genetic condition called Med13L syndrome. MED13L syndrome is an autosomal dominant condition due to pathogenic changes in the MED13L gene which is located on chromosome 12. This intellectual disability ranges from moderate to severe. All individuals have developmental delays with speech being most prominently affected.

Although Charlotte is not autistic or display behavioural problems associated with this diagnosis, it does now explain her severe speech and language difficulties, her poor eyesight, gross motor delays and her right foot that turns inwards. It also explains her intellectual disability and delays. Charlotte also has upper frequency deafness and wears hearing aids.

There is no research or information available as to how much to expect from Charlotte in terms of achievements so ongoing speech and language therapy is crucial to ensure she reaches her full potential.

From the age of 3 years old Charlotte would receive a six-week block of Speech and Language therapy sessions every couple of months. We could see that the local provision and support Charlotte was receiving was not adequate or consistent enough to meet Charlotte's needs. At each block of sessions we would be allocated a new Speech and Language therapist. It would take the new therapists a few sessions to get to know Charlotte and before we were started to make progress the therapy sessions would be over.

When Charlotte started school the speech and language therapist allocated to the setting would see her once a fortnight for 30 minutes. Speech and Language was Charlottes main area of difficulty and since she wasn't receiving the care and support she needed my husband and I sought to find a private speech and language therapist who would come to the house to work with Charlotte. We chose Emily because she is a very professional paediatric speech and language therapist and has extensive experience of working with children with complex needs. It was honestly the best decision we have made.

From day one Emily was fantastic with Charlotte. She has a natural, fun and enthusiastic manner with children and instantly built a great relationship with our daughter. Each week Emily creates fun,

interesting and interactive activities within her therapy sessions keeping Charlotte engaged and motivated, developing Charlotte's speech without her even realising it's a therapy session.

We are also delighted to report that Charlotte has made astounding progress in the time Emily has worked with her. When Emily started therapy sessions with our daughter, Charlotte would use only 2 words. Emily has taught us all Makaton and Charlotte now speaks in sentences. This has made such a difference to us as a family, we can now understand what Charlotte is saying and what she wants. It's also given Charlotte more confidence to play with friends and actively participate in her classroom. Emily has now progressed into incorporating reading and writing skills into Charlotte's sessions. We are amazed at the progress Charlotte has made, especially with her diagnosis of MED13L Syndrome.

With Emily supporting Charlotte, we still liaise with the speech and language therapist allocated to the school, however, there is no doubt in our minds that the consistency, support and therapy that Emily provides is by far superior.

We cannot thank Emily enough for her continued dedication, commitment and the support she gives Charlotte and to us as a family. We cannot recommend her services highly enough.