

GEORGIA: RUSSELL SILVER SYNDROME

HER STORY SO FAR

Problems in the pregnancy began at 17 weeks following an amniocentesis test. The amniocentesis didn't go according to plan and a couple of hours later I was back in hospital because it seemed that the membranes had ruptured. The doctors were very pessimistic and I was put on bed rest for a few days and monitored closely. Following numerous checks and scans I was discharged from hospital after two weeks and told to take it easy for the rest of the pregnancy.

From then on I had to attend the antenatal clinic three times each week. I was monitored each time and also had to have blood taken and various other tests. I also had ultra sound scans each week from around 22 weeks. It was at about this time that they discovered that Georgia wasn't growing as well as she should have been. At 28 weeks and following another scan and monitoring it was decided the baby was not doing at all well. The Consultant though, really wanted the pregnancy to continue for as long as possible on the basis that the baby would be better off in the womb than in an incubator. This continued for another 8 weeks by which time the doctors decided hopefully now the baby would be strong enough to enter the world. They estimated at this stage that Georgia was about 2kg. She was also breech so they decided a caesarean section would be advisable.

Georgia was born at 36 ½ weeks gestation on the 10th September 1997 weighing just 1.71 kg (3lb 12oz). She was immediately whisked off to the Special Care Baby Unit (SCBU).

The first few days were very difficult. Georgia was breathing with the aid of oxygen and when I first went to see her she was so tiny and covered with all sorts of tubes. I was allowed to hold her for the first time after about 24 hours.

Georgia was a bit of a puzzle to the medical staff because they couldn't understand why she was so small. They were convinced that she was only around 32 weeks but we knew this wasn't the case. After a few days she was breathing unaided but she was very reluctant to feed. This continued to be a problem for some time and was the only thing after a couple of weeks that was really keeping her in SCBU. We tried everything to get her to feed, but she was taking very little orally. Finally though, after 3 weeks and 6 days she was discharged.



Georgia a couple of weeks old

The feeding problems continued at home and Georgia was very reluctant to feed. It took us over an hour to get her to have about 20ml and by the time she had finished it was almost time for the next feed! During those first few days we tried so many different teats but she just wasn't interested. She never woke in the night for her feeds so we had to set the alarm clock so that we could wake her up. After 8 days at home the midwife was not happy with her lack of appetite and she was also losing weight. She was readmitted to hospital overnight for observation. During this time the hospital staff apparently had no problems feeding her so she was discharged the following day with the disposable teats that they were using. We had found that Georgia did go through periods of feeding reasonably well which was obviously what had happened in hospital. At home we were back in the same situation; she still took hours to feed; she just had no interest at all.

We struggled along for another week or so and then Georgia became ill with a cold so the feeding was even more difficult. After a couple of days we took her to the doctors because we were worried about her breathing. He checked her over and didn't feel it would be too much of a problem but he was quite concerned about her feeding difficulties. During the night her condition deteriorated and she seemed to

be having a lot of difficulty breathing. The following day Georgia was readmitted to hospital with suspected bronchiolitis.

It seemed to take forever for the hospital to sort her out. They gave Georgia numerous tests including a lumbar puncture because it was suspected she may have had meningitis. At about 2am, after hours of waiting, she was diagnosed with Respiratory Syncytial Virus (RSV). The probability was that she had caught this virus when she was in hospital overnight because of weight loss and feeding difficulties.

The next few days were a very difficult time for us all. Georgia was back in an incubator, on oxygen and being sustained intravenously. She had one to one nursing care and at one point she was prepared to be transferred to ITU at the Birmingham Children's Hospital.



3 Months Old

One doctor was very concerned about her condition; she asked 'why is this child so emaciated?' It was during this stay in hospital that 'IUGR' was referred to. I had heard this mentioned a couple of times and informed by a nurse the letters stood for Intrauterine Growth Retardation. At this stage we didn't realise how much this was going to be part of our lives. After a few days Georgia's condition began to improve slightly so she was taken off the oxygen. She also had a Nasal Gastric Feeding tube placed to help her because she was too weak to feed normally (this is a tube that runs from her nose to her tummy). Georgia had the tube for a few weeks, when it was removed once again she would not feed. After a few days it was suggested that Georgia was well enough to go home. Although we were desperate for her to come home we were very concerned that nobody was really taking us seriously. We asked if Georgia could stay in until the next day to see if she would gain any weight. Georgia stayed in hospital

and was weighed the next day; yet again she had lost weight. They decided to keep her in a little longer.

Georgia had lots more tests in an attempt to find out a reason for her size and lack of appetite. This included a 'sweat test' to rule out Cystic Fibrosis. Finally, after 3 ½ weeks in hospital, although her feeding remained a problem she came home under the care of the community paediatric nurse. The nurse would come to visit us at home on a regular basis and monitor Georgia's weight and feeding. We were still no closer to finding out why she was the way she was.

At home the problems continued and after one week Georgia continued to lose weight. It was decided to place a nasal gastric tube on a long term basis. Although whilst in hospital we were so desperate for her to feed normally we were now resigned to the fact that she needed extra help, we were relieved because it took a lot of pressure off us and we hoped that this would help Georgia 'thrive'. We plodded along for the next couple of months, Georgia had another couple of admissions to hospital with coughs, colds etc. She nearly spent her first Christmas in hospital. Fortunately, she was discharged on Christmas Eve.



Georgia at 9 Months Old

By early February we had had enough. Georgia was now totally tube fed and although she was gaining weight it was very slow and she would often vomit after the whole feed. She had now been labelled 'failure to thrive'. Her development was also much delayed. She wasn't reaching any of her milestones; for example she didn't smile until she was about four months old. She was seen by the paediatrician at our local hospital every couple of weeks, but we felt that we were not getting anywhere. He kept saying 'she will catch up eventually'. We didn't agree and felt

Georgia was not receiving the specialist investigation and care she deserved.

In desperation we took her to the A & E department at the Birmingham Children's Hospital. She was admitted immediately for tests. Georgia was seen by many specialists including a neurologist who painted a very bleak picture. He didn't think she would ever walk or talk, and would be significantly retarded in many ways. She was also seen by a geneticist who at this time didn't make any diagnosis but said that he would see her again in a few months. Unfortunately, within a few days Georgia became infected with Rota virus (a very severe gastroenteritis). She was extremely ill for a few days and lost over ½ Kg in weight in just 24 hours. Luckily once the virus was out of her system she managed to regain the lost weight. To our dismay after about 3 days she came down with pneumonia. This of course knocked us back even further. She stayed in hospital for about 2 weeks and we were very happy to take her home (even though we were still no closer to a diagnosis) just in case she became infected with something else. But at least we now felt that she was under specialist care being a patient of the Birmingham Children's hospital.

We tried to carry on with a normal life although this was generally quite difficult. Georgia continued to get many infections and the vomiting also remained a big problem. Most mornings she would vomit, usually when I was about to take her sisters, Jessica 6 and Chloe 2 years old, to school.

On Georgia's first birthday she weighed 4.9Kg (10lb 11oz) and she was 63cm in length.



Georgia's 1st Birthday

When Georgia was 14 months old she was once again admitted to the Birmingham Children's Hospital for more tests. It was during this stay in hospital that she was visited again by the geneticist. He told us that when he saw Georgia previously he suspected that she may have Russell Silver syndrome but at the

time Georgia didn't display enough of the characteristics of this condition, so was reluctant to give a firm diagnosis. Now he was very sure that she had got RSS. We were relieved, although we were very concerned that she had this condition at least we now knew the reason why she was so small and had no appetite. We also had the hope that she would improve over the years.

We could now give Georgia the help and support that she needed. Every time we went to the doctors or for a hospital visit we would go armed with the booklet from the Child Growth Foundation. We learned so much about the condition over those first few months and it was wonderful to talk to other parents who were going through the same situation or had come through the other end.

Georgia continued to gain weight very slowly and she continued to be very reluctant to take anything orally.



Georgia at two years old

One of the problems with Russell Silver syndrome can be Hypoglycaemia; we learned about this the hard way. One morning we tried to wake Georgia but she was very lethargic and reluctant to wake up. At first we thought she was just tired but it became apparent that her feeding tube had somehow become dislodged during the night. We didn't know when this had happened so was unsure how long she had gone without any nutrition. We didn't know what to do, so we first of all tried to get hold of Georgia's nurse without success. We then tried her consultant and again this was unsuccessful. Eventually his secretary managed to track him down and instructed us to call 999 immediately. The whole thing was a bit of a daze really and I didn't know what I was doing. It really was a nightmare and the worst of it was that it was totally out of my control. By the time the ambulance arrived Georgia was also fitting and making some awful noises. The paramedics thankfully took control and she was rushed straight to

A & E. Her consultant met us there and we knew she was in safe hands.

It turned out that Georgia had become hypoglycaemic and had gone into a hypoglycaemic coma. Her blood sugar levels were so low they were measuring zero. She was administered the necessary medication and showed a little improvement. She was admitted to the ward some time later but it took about 24 hours for her to come around. Upon being discharged from hospital three days later and we were given equipment to check her blood sugar levels in case of a similar problem. Thankfully, this has not happened since although we are always aware, just in case.

When Georgia was 2 ½ she was fitted with a gastrostomy (a feeding tube directly into her stomach through a hole in her abdomen). At about the same time she started at a nursery for physically disabled children - at this stage Georgia was still not walking. Our aim and hope was for Georgia to attend the Special School, to help her development, with the objective of her progressing to the local mainstream school.

It was probably at about this time that we felt we could 'see the wood through the trees'. Georgia seemed to be much healthier and not so 'delicate'. She started to walk on the 1st December 2000, at three years and nearly three months old. We were all so proud of her and she took the lead role as a 'star' in the schools' Nativity where she took a few furtive steps.



Georgia starting to walk

Her talking was still a big problem though, and although she had regular speech therapy we could see no real improvement.

At 3 ½ years old Georgia began growth hormone therapy. At this time she still only weighed just over 8kgs and her height was 78cms.

Within a couple of months she had grown considerably and her appetite had increased. Georgia seemed a different 'little' girl. After six months on growth hormone she was 85cms. That really was probably the turning point for Georgia. She became much more physically able and her speech was coming along slowly.

Our hopes were realised when Georgia started mainstream school in the reception class with 20 hours support from a teaching assistant to help with her educational and medical needs.



Georgia on her 7th Birthday

Georgia has continued to progress really well, at the age of 7 years and 2 months she weighs just a little less than 20kg and is 115cm tall. Her growth has been quite dramatic and currently she is having a break from growth hormone, we will see what the impact of that is. She came off growth hormone 8 months ago and so far although her growth rate has dropped back a little she is still growing. Georgia is also doing very well in school and works incredibly hard. She is very popular at school and has lots of friends. She has just been chosen to learn to play the cello, this is quite an achievement because only four children out of a total of 90 get the chance. She loves dancing, horse riding and swimming.

Seven years ago we would never have dreamed that Georgia would have achieved so much. She tries so hard and we are all so proud of her. Georgia is an absolute delight and we wouldn't want her any other way.

Written by Jenny Child, Sutton Coldfield, England
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