

DEDICATION TO A SPECIAL PERSON—AMY



It was Wednesday, August 28th 1991, when little angel Amy came into this world in Liverpool Women's Hospital, UK. She was born 8-weeks prematurely weighing merely 2 lbs 11 oz. She was declared to have an intrauterine growth failure.

Here is what Amy's mum has to say about her birth: "Looking back, we think that it was apparent to specialists at her birth that there was a problem—silence when she was born, her placenta was thin and transparent. Despite the fact that a geneticist was brought to see her, nothing was said to us, and we presumed that this was a normal birth".

In the beginning Amy met all her milestones; she was tiny yet an active baby. She walked at 13 months, talked

on time and health visitors told Amy's parents that she would need to wear diving boots to slow her down. At around 18 months, Amy visited the Genetics Department of the Alder Hey Children's Hospital, Liverpool, UK for follow-up, where it was advised that something was seriously wrong with her—something very rare—and that, most likely, she wouldn't grow—they gave it a name—'Amy syndrome'. One of several disorders was suspected and tests were done, but no specific diagnosis was decided upon.

Amy continued to thrive in some ways but not in stature. She was a nightmare to feed but remained happy and full of energy. She began attending a mainstream school but couldn't quite keep up, so was transferred to a Moderate Learning Difficulties School where she made lots of friends and loved her work. When Amy was 5 years-old, her parents, while researching on the internet, came across a rare genetic disorder of DNA repair deficiency called Cockayne syndrome (CS). The clinical features of this disease and photos of CS children indicated that Amy might be suffering from this syndrome. Tests were carried out in the UK but no concrete results came back. Yet her parents strongly believed that Amy had CS.

Around the age of 11, Amy began to deteriorate. Firstly she developed a tremor and then her balance began to fail. She fell over regularly and began walking with a very poor posture. She kept asking her mother "What is happening to me?" And she began to feel very sad. Despite regular visits to a number of doctors, they could give her very little help. At this stage Amy's parents took the challenge to ask "What exactly was wrong with Amy?"

Amy was 14 when her parents raised enough money to fly to the USA to meet Dr. Edward Neilan at Boston Children's Hospital, who was studying CS at that time. Dr Neilan carried out genetic analyses on Amy's chromosomes and found a mutation, which indicated (together with her symptoms and appearance) that she had atypical CS. When the diagnosis was confirmed, Amy's mother said "As a mother, I had known this for nearly 10 years and, in one way, my mind was at peace with this news but, in another, I was broken-hearted".

At this time Amy started suffering from tremors. Moreover, she was unable to eat, drink, dress or carry out her everyday tasks without help.

Subsequently Amy was taken to a neurologist, Dr. Peter Kang, also in Boston, who prescribed medications for her tremors. Within 10 days Amy's tremor had essentially disappeared and this distraught girl was back to her former happy self. Not only that, but a number of other CS children began taking the same medication and they too regained some measure of dignity. From then on Amy became desperate to help other children by participating in several invasive tests, carried out at experimental levels, and by saying "If it helps others then I want these done".

Back in England Amy's parents' thinking was that they did not want any parent or child to ever feel as lonely or isolated as they and Amy had. So in 2007 they set up a charity organization called "Amy and Friends (Cockayne Syndrome Support)", and since then have united and helped over 50 families from across the world suffering from CS. This charity can be found at the website www.amyandfriends.org (Cockayne syndrome support).

Through this organization Amy and Amy's parents have brought together a large number of children suffering from CS, provide them happiness, love and support, while sharing together heart-breaking moments when children as young as 20 months and as old as 20 years lose their life. Brave Amy has visited several of her dying friends and helped them on their way to heaven by saying "Go now, our friends are waiting to take you, don't stay here—go and run and play".

She tells everyone she meets that she is glad to have CS so that she can help others. She is a pleasure to be around, brings love to those who meet her and once anyone meets Amy, she is never forgotten. Her teacher once said that what Amy lacks in stature she makes up in spirit.

Amy is now suffering from an underactive thyroid, kidney failure, hypertension, diabetes, high cholesterol, raised liver enzymes, a low grade glioma on her thalamus, kyphosis, lordosis, scoliosis and is stiff and in regular pain. She is unable to do most things she loves doing and very often feels lonely, despite having so many people around who love her. She lives now every day wanting to live her life to the fullest and dreaming of the day when her body is free from this illness.

Never before in my life I met a person who has been suffering so much, yet very bravely faces the situation and at the same time showers her help, love and affections to others suffering as her. She must be one of the bravest persons known. She has touched my heart so much in such a short time that, not only have I decided to work for this charity organization, but also dedicate this book to her. I gave her an Indian name—DIYA—which means a 'clay lamp' that burns to give light to others.

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