Jessika Wynter Anastasia Parry

DOB: January 28th 2010

Diagnosed with type 1 SMA on September 30th 2010.

SMA (Spinal Muscular Atrophy) is a genetic motor neuron disease. One in 40 people is a carrier, meaning that they have a defective or deleted copy of the SMN1 gene. Two carriers have a 25% chance of producing a child with the disease. Severity and age of onset vary considerably. Type 1 is usually diagnosed before 6 months of age and the child will never develop the ability to sit, type 2 is typically diagnosed before 12 months of age and the child will never develop the ability to walk. Type 3s are diagnosed as late as in their teens and are marked by loss of ability to walk, type 4s usually are not affected until adulthood, around the age of 35.

Type 1 is the number one killer of children under the age of 2. The disease affects the development of muscles; those used every day for sitting, crawling, walking, swallowing, breathing, grasping objects, lifting a head, etc. 1 in 6000 infants and children, of all races, genders, and social classes, is affected (21,000 worldwide). 50% of those diagnosed before the age of 2 will die before their second birthdays; 13,000 this year alone.

After hearing all that one may well lose hope but hope exists, there are protocols that have been developed that can and will make a big difference in the life of a child with SMA. The NIV (Non-InVasive) respiratory protocol, which uses a group of techniques and machines to aid in development and exercise of respiratory muscles, has been developed and taught worldwide by Drs Bach, Swoboda, Schroth, as well as many others. Using these protocols can extend the lives of these children considerably by keeping the respiratory system strong and able to survive respiratory infections that would kill the child otherwise. The protocol uses these machines; a Bi-level Positive airway pressure (BiPap) machine to aid in night-time breathing and expanding the lungs more than the child could on their own, an in-exsufflator (cough assist) device to simulate a real cough to clear secretions from the lungs and also to aid in expansion, a suction machine will remove excess secretions that the child cannot manage on their own and a pulse-oximeter will monitor the child’s oxygen saturation, notifying the parents of oncoming respiratory distress. Chest physiotherapy includes clapping, with a cupped hand, on the child’s chest, sides and back, around the rib cage, to loosen secretions in the lungs; using a modified ambu-bag to expand the lungs and laying the child at a downward angle on a 30’ wedge to drain secretions. Placing a G-tube, for feeding, as early as possible may also prevent complications from having surgery after the child can no longer tolerate intubation (a tube is inserted into the lungs during surgery to prevent the child from going into respiratory distress).

Even though these children have almost no motor abilities, their brains still function and develop normally. This means that they still require to be taught, to be stimulated, to be entertained and played with. Of course they also need copious amounts of love and care, but that goes without saying. Thankfully technology has progressed to the point where we can easily meet the needs of these children; there exist computer/communication systems that are designed to be operated simply by using the eyes (the eye-gaze system), there are voice amplification devices that can be used to augment the weak voices of these children, helping them to develop their personalities and sense of self; iPads can be used to play games and communicate using it’s sensitive touch screen and specialised apps, some parents (including ourselves) have built multipurpose stands and frames that can be used for hanging toys, positioning portable DVD players, etc. There are many sensory toys available over the internet that feature lights and sound that can simultaneously entertain, stimulate and relax. Other toys can be modified to make them easier to use; switches can be replaced with ones that are easier for these kids to manipulate, ultra sensitive touch switches, laser switches, etc.

Water therapy is an excellent tool since movement is much easier when in such a buoyant environment. The main thing to consider is that there is an increased risk of aspiration if the child swallows water so flotation devices of some type can be used, around the neck, to keep the head above water.

Transportation also becomes an issue as parents decide whether to choose to transport their children in wheelchairs or in car beds; vests have been created that strap onto the child and then strap into two seat belts on a rear car seat so that children can be transported laying down. Vehicles will need to be adapted for wheelchair transport; ramps installed and tie downs provided as well as power options for transporting and using equipment during car travel.

These children will need positioning equipment for seating, standing frames for standing (weight bearing on the legs helps develop bone density, preventing fractures), orthotics to prevent contractures; parents will need to be resourceful, creative and persistent to be able to provide everything that their children need and deserve to maintain a suitable quality of life for their children, for these children who have to fight so hard for each and every breath and deserve so much to have a fighting chance while the medical world struggles to find a cure for this terrible disease.