Arm in ARM - sometimes we can't stop the waves, but we can learn to swim

by Kate Tyler

In September, I had the immense good fortune of attending the Max's Trust Annual Conference, an inspiring and uplifting weekend event.

The charity, Max's Trust, is named after a beautiful boy named Max Finnigan, who was born with anorectal malformation (ARM). This diagnosis meant that Max needed emergency surgery to make a life-saving stoma at 24 hours of age, followed by further specialist surgery (PSARP) at 11 weeks and a reversal of his stoma at 8 months of age. Tragically, at 17 months, Max died very suddenly and unexpectedly from something unrelated to the ARM. His short life was filled with love and happiness and it is in memory of their adorable son, that Cassie and Jon Finnigan set up Max's Trust to 'Provide Support and Information to the UK Anorectal Community'.

Max's Trust has many aims in common with TOFS Charity, such as: raising awareness, sharing information, supporting families and adults affected by ARM, working in partnership with health professionals and supporting research initiatives. They are dedicated to inspiring the ARM community of the UK.

ARM is the term used for a group of congenital malformations involving the bowel and anus, where they have connected to the urinary tract or, in female infants, to the vagina. These also have an impact on the pelvic floor nerve and muscle functioning, resulting in health challenges that may present themselves in childhood and later in adulthood. For those infants born with ARM, most will require surgery to position the rectum and anus and, if connected, to separate the bowel from the urinary or reproductive organs.

Paediatric colorectal surgeon Dr. Alberto Pena introduced the posterior sagittal anorectoplasty (PSARP) in 1980, a procedure that has transformed the treatment for many born with ARM. There are different classifications of ARM; this means that some infants will not need a diverting stoma and the surgeon can proceed to the repair. For others, surgery is needed in the first days of their lives in order to remove waste from their bowels. Repair for these babies means three operations: formation of a stoma, repair surgery and finally reversal of the stoma if, and when, the family and surgeon decide it is the best time to do so.

In addition to being born with OA/TOF, I too was born with an anorectal malformation where the lower part of my bowel had not formed correctly and stool could not pass through my body – my digestive system was in effect blocked. My life saving surgeries, for both OA/TOF and ARM were at just a few hours of age - 65 years ago.

Three or more of these anatomical differences result in a diagnosis of VACTERL Association. The acronym refers to an interruption in development in the very early stages of pregnancy, resulting in Vertebral, Anal, Cardiac, Tracheal, Esophageal, (American spelling) Renal and Limb defects.

VACTERL occurs in around 1:10,000 - 1:40,000 births.

As with OA/TOF repair, it is essential that the complex surgery for ARM is performed by skilled, experienced paediatric surgeons, working within a multidisciplinary team of health-care professionals. As babies and their parents embark on this journey it is vital that they are provided with information and support to help them navigate what can be a very distressing and challenging pathway. Those affected by this rare anomaly require a lifelong approach to their healthcare in order to achieve the best health outcomes. This includes collaborative working at all stages and planned transition from teenage years to adult care is particularly important. Local health professionals must be able to signpost and refer adults to the specialist adult colorectal, urology and reproductive specialists they may need in later life. Ongoing psychological support is also vital for those who are affected by invasive or frightening medical experiences of childhood.

l-r, Cassie Finnigan, Helen Millard, Jon Finnigan, Kate Tyler and Lauren Stahly

Throughout the weekend, the conference programme was stimulating and informative, providing education and advice. Paediatric surgeons Joe Curry, Govind Murthi, Clare Skerritt, Jonathan Sutcliffe, Bala Eradi and Adult Surgeon, Dermot Burke, discussed the principles of reconstruction surgery, raising awareness of missed diagnosis, the physiology of bowel management and the importance of supporting research to help shape future care.

Caroline Gainsbury and Julie-Ann Milbery, specialist nurses from Great Ormond Street Hospital, facilitated workshops around stoma care, toilet training and preparing for nursery and school. There were many opportunities to ask questions and engage with all the health professionals. Particularly valuable for me was the informal chat with other adults born with ARM, followed by conversations with Jonathon Sutcliffe and Dermot Burke, both of whom were very interested in the different challenges we adults had encountered. It was a one to one consultation with UK experts and it was very much appreciated by those present.

We watched a fascinating webinar entitled 'Building Resilience for the Harder Times', presented by Brian Marien, demonstrating the value of being in tune with our emotional barometers and accepting that life can be very challenging and that, "sometimes we can't stop the waves, but we can learn to swim."

Finally, we listened to a deeply moving talk from Lauren, a young adult born with ARM, who shared her journey with us. At this point, many people were reaching for the tissues - I am sure parents at the conference will cherish her inspiring words.

For me, the conference was far more than learning about my anomaly and how differently it is managed in the 21st century. It was about the coming together of children, their parents, adults born with ARM and the professionals who care enough to give up their time to support us all, to listen to our concerns and value our experiences. I think the adults present really felt that we too were being listened to and could help shape future care.

Although the number of infants born with OA/TOF and additional anomalies is small it is, in my opinion, essential to reach out to all with rare and complex congenital diseases. The importance of lifelong support and care for those with OA/TOF and ARM is vital and hopefully, together, we can influence future service provision and make everyone's journey a little easier.