

How did life change during a worldwide pandemic? The story of a little boy with WAGR syndrome.

In the UK, shielding measures mean that people in a clinically extremely vulnerable group are advised by the government to stay at home and avoid non-essential face to face contact with other people [1]. In other countries, although perhaps not directly recommended by the government, many families choose to go into voluntary shielding. I am very grateful to be given the opportunity to connect with one such family. Sarah and I spoke about her experience quarantining at home in Belgium with her 4-year-old child - Logan (names of mother and child changed to protect identities). Logan, a little boy who is truly one in a million, was diagnosed with a rare genetic condition called WAGR syndrome at the age of 3 months.

WAGR syndrome is caused by an autosomal dominant deletion of a region in chromosome 11 [2]. The cardinal features of the syndrome correspond to each letter of the syndrome's name. 'W' stands for Wilm's tumour, a form of kidney cancer caused by the abnormal development of immature nephroblasts. Logan has up to 77% chance of developing a Wilm's tumour [2] and thus, he has regular ultrasound monitoring of his kidneys every 3 months.

The 'A' in WAGR stands for aniridia, describing the absence of the iris of the eye. When Logan first opened his eyes to the world, a specialist noticed that the coloured part of his eye was missing. This has been linked to photophobia, nystagmus (involuntary movements of the eye) and impaired visual acuity as well as development of cataracts, glaucoma and ocular surface disease [2].

The 'G' in WAGR refers to genitourinary tract problems, caused by a deletion of the *WT1* gene [3]. *WT1* codes for proteins involved in the structural development of the genitourinary tract. In Logan, this manifested as hypospadias – where the opening of his urethra did not develop in the correct location – cryptorchidism (also known as undescended testicles).

Finally, the 'R' in WAGR stands for a range of developmental abnormalities. This can vary significantly between individuals, but intellectual disability (IQ<74) is recorded in 70% of patients. Other developmental disorders such as ADHD and Autism spectrum disorder may also be present [2]. Logan is diagnosed with autism and sensory processing disorders (SPD).

With WAGR syndrome, Logan has to see a multitude of different specialists. From the ophthalmology team for his aniridia to a urology team for his hypospadias to a nephrologist for the monitoring of his kidneys, there are many specialists involved in his care who need to communicate effectively amongst themselves as well as communicate with his parents.

Worries of compromised care

In our phone conversation, Sarah said she very grateful to be attached to a multidisciplinary team at a large teaching hospital. They have regular meetings to discuss Logan's care and she is happy that they have left no stone unturned in making sure that all avenues of treatment were explored. However, with the COVID-19 pandemic, there have been worries that the care of her child would be compromised.

For example, after receiving a string of notices informing her of the cancellation of many other appointments, Sarah was quite worried that the quarterly observational ultrasound for a Wilm's tumour in Logan's kidneys would be cancelled. She mentioned that she was looking private healthcare alternatives but was unsure if the assessment would be up to par with his regular ultrasounds as the private specialists would not have access to previous ultrasounds. This is a valid worry because if a Wilm's tumour is found in an earlier stage, the treatment comorbidities significantly decrease [4]. Similarly, the EURORDIS Rare Barometer Covid-19 experience survey found

that interruption of routine rare condition healthcare has been detrimental for 66% of survey participants [5].

Thankfully, Logan's ultrasound appointment was not cancelled. Nonetheless, this does not mean it cannot happen to another patient - there are still many rare disease patients who are struggling to get their concerns noticed because it is "not considered an emergency". As the priorities of healthcare systems change with the pandemic, the health of those with chronic rare conditions requiring routine care seems to be taking the hit.

Why is it important to consider all parts of the patient's life?

Although having uncomplicated WAGR syndrome means that Logan would not be considered as part of the clinically extremely vulnerable group, he also has an associated immunodeficiency which requires regular prophylactic antibiotics. After experiencing several spells of pneumonia which got him hospitalised for the past few years, Sarah decided it would be best for Logan to shield. For this, she decided to take a break from her job to stay at home and look after Logan full time.

With schools plus other respite facilities being closed, it fell on Sarah to keep up with Logan's daily therapy and medications. Logan's autism and SPD diagnosis means that he also frequently has emotional outbursts as a result of overstimulation. It must have been very stressful for a 4-year boy with autism to suddenly adjust to a whole new routine with no explanation as to why compounded by being unable to communicate verbally.

As the months continued, Sarah described how Logan's emotional outbursts progressively escalated to become more extreme. The medication for his sensory overstimulation was not working effectively and the paediatric psychiatrists they contacted either refused to see them or put them on a waiting list due to COVID-19. The reduced availability of care services and managing Logan's outbursts 24/7 weighed down hard on her and her husband. In a facebook post about the subject on a page she created for her son's journey, she says it "became too much". The stress and frustration of not being able to get the best for her son alongside feeling incredibly tired herself without any breathing space would have been incredibly challenging to manage. Primary caregivers can experience fatigue, frustration, guilt, and loneliness leading to a very real burnout – which can be detrimental for their own health as well as the health of the person they are caring for [7]. It is important for structures to be in place which allows for caregivers to have some respite for themselves in order to prevent burnout.

I am glad to say that Sarah and her husband managed to get some respite after the doctors picked up on their weariness. In an MDT meeting, everything was discussed to be medically great for Logan – there were no signs of a Wilm's tumour and successful surgeries for his hypospadias mean that he has a functional urinating capacity. The team could have just stopped there with an "All's good!". But a simple question asking, "how are you doing at home?" revealed all of the challenges that Sarah and Logan were facing. In another scenario, the team could have taken the easy route and said that there were not any options available for Sarah because of COVID-19.

Even when it seemed that all options have been exhausted, the team endeavoured to find alternatives and it was fruitful. They tried their best to find respite for the family and after pulling a lot of strings, they were able to secure a place for Logan in an observational care facility where they are currently observing his sensory outbursts with the aim of developing long-term therapies. The 3-month admission to the facility also gives an opportunity to Sarah and her husband to recuperate.

By considering the psychological wellbeing of both Logan and Sarah, the doctors provided good holistic care which did not just focus on the medical minutiae of WAGR syndrome but his overall individual needs (which are so interconnected to the health and wellbeing of his mother as his primary caregiver too).

In a pandemic which has disrupted the care of so many rare disease patients, this is a story of positivity. Even if consultations have become remote, there is a lot of power in providing holistic care and maintaining a clear dialogue which allows patients to express all of their concerns. A holistic approach becomes all the more important when caring for a complex rare disease patient, like Logan. Through Sarah and Logan's story, I have been inspired to never compromise on patient care and always trying my best regardless of the situation. It is possible in the middle of worldwide pandemic, so it should be possible at all other times too!

References

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