

Ezras Spinraza Journey

My son Ezra is 9 years old. Born in 2016, he was one of the first children to be treated with Spinraza in England on the EAP, he was diagnosed and treated too late at 5 months old. Consequently, he lives with a range of complex needs.

By just 5 weeks old Ezra was in intensive care with a common cold. He has a very severe form of the condition; without treatment and intervention he would have died before his first birthday. If treatment stopped now, he would rapidly deteriorate and would probably not see his teenage years.

Ezra is an intelligent young man with a sense of humour beyond his years. He attends mainstream Primary school in his powered wheelchair and is meeting above expected levels across the curriculum. He is confident, social and outgoing.

As a baby, Ezra made significant motor gains following treatment. At the age of 1 he lifted and held up the weight of his own head for the first time, at the age of 2 he sat independently, his assessment scores continued to show significant progress. His respiratory strength improved and by the age of 3 he was able to manage colds at home without hospital admissions.

Now that he is bigger and heavier, his Hammersmith assessment scores have plateaued. He has not made any officially recorded 'gains' in his assessment score over the last few years. However, the results of standardised clinical assessments that you see in the data reports do not reflect the real world gains we see at home.

The increase in diction and volume of his voice enables him to communicate without interpretation- he can play freely with his friends and participate wholly in class discussions. This shows huge improvements in his facial and bulbar function as well as his respiratory drive. He can now shout so loudly that I can hear him from the bottom of the garden, enabling him to play independently, and enabling me to be able to safely leave him to it. It is a development that has improved both his independence and mine.

Now 9, Ezra is learning his own limits, and we are cautiously following his advice. Last summer he decided for the first time that he could sit up in his car seat for an extended period of time, managing a 6-hour journey across France. Not long ago he would have to lie flat across the backseat with his bi-pap ventilator and regular suctioning.

Now, not only can the whole family actually fit in the van -family days out and holidays are far less stressful. The times carrying a suction machine, cough assist and ventilator around with us everywhere we went are long gone.

Ezra has not eaten orally since he failed a swallow safety test at just 5 months old. With no updated clinical guidelines post treatment - there has been no monitoring of his swallow since. But Ezra is determined and wants to eat. He now regularly asks for dinner to be blended so he can join us at mealtimes and manages, with great determination, to swallow several teaspoonfuls.

Ezra has an incredible mastery of his iPad, he independently searches for what he wants to watch, learn or play – giving his sister and his dad a run for their money on Roblox games! His fine motor skills have enabled him to be able to fully access the school curriculum – using his iPad to write the most wonderfully imaginative stories.

I have many other anecdotes of how seemingly small, clinically unmeasured gains have improved Ezra's health and wellbeing, and the wellbeing of the whole family in a huge way. Spinraza has not only kept Ezra alive, but it has also enabled him to thrive.

So why Spinraza ? Why have we not switched treatments as others became available? Ezra was too heavy to be a safe candidate for Zolgensma –and to be honest, at the time, I was suspicious of a 'one and done' treatment with no long-term evidence. I am reassured by the Spinraza 'top-ups' and I'm sure he has a boost in energy and strength immediately after the infusion.

Why do we choose to let Ezra have 3 intrathecal infusions a year, why wouldn't we switch to risdiplam, with its much less invasive method of administration. I have several reasons for our personal choice;

We know it works – real world evidence shows that both treatments have very individualised outcomes. I know Spinraza works for Ezra – I cannot risk any level of deterioration. A small loss could stop him from being able to drive his powered wheelchair or put him back into hospital with his next cold.

It goes straight to where it is needed. Ezra has a very severe form of the condition. It gives me peace of mind to know that the medicine is going straight to the source of the problem.

It's not a daily medication that I am responsible for. It suits us to only think about his medication 3 times a year – for the rest of the time we are free without the responsibility of administering a daily, hugely expensive, medicine. That is a responsibility I am very happy to hand to a trained clinician!

He tolerates it well. Ezra understands why he has his 'muscle juice', he only requires a numbing cream and remains remarkably calm throughout the procedure. There are never any tears, and he shows no signs of procedural anxiety. We stay in the hospital for just an hour afterwards for monitoring, then we go home. It also presents the useful opportunity to see other members of Ezras multidisciplinary clinical team.

Not only does everyone respond to SMA treatment in different ways, but people also have different priorities. This is why it is essential that we retain a choice of SMA treatments in the UK. There is not a one fits all solution to SMN targeting therapies.



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