Rewriting the future of how Scotland cares for dying children

A brief checklist from the Batten Disease Family Association (BDFA)

BATTEN DISEASE FAMILY ASSOCIATION
Together we WILL make a difference
Registered Charity in England and Wales 1205650

BDFA has supported Inverness parent Donna and her son Reece throughout his journey with CLN2 Batten disease, Reece died in May 2024 at Rachel House. BDFA continues to support Donna, and we thank her for supporting us to chronicle the key milestones along their Batten Journey.

Reece's Batten Journey

1 The diagnostic odyssey

Donna first had concerns about Reece age 2, his speech was not developing and he was referred to a Speech and Language Therapist, he had his first seizure age 3, his behaviour rapidly changed and he was diagnosed with autism. Donna knew there was something else happening to Reece, he was stumbling and falling daily. His vision was poor, and he stopped progressing. A year later he had a sight test, and the diagnosis of CLN2 Batten disease came shortly after. The timeline between initial symptoms and diagnosis needs to improve – Parents need to be listened to when they have evidence of changes in their children, the wait for diagnosis impacts on family wellbeing for years to come.

Children with CLN2 can be offered treatment to slow the disease and early access to this is vital.

2 The diagnostic consultation

Donna phoned the consultant to find out whether there was any news from the blood tests. He said, 'Reece will be diagnosed with Batten disease, but I didn't want to be the one to tell you, you'll have a diagnostic consultation in 2 weeks with the metabolic specialist.'

When she went to the consultation, she was met by the Lead consultant and 6 other members of the clinical team, this is not uncommon, often there are more people in the room. There must be an understanding of the grief for parents facing such a diagnosis and the feeling of scrutiny. Numbers of professionals should be limited to 2 or 3.

Parents should never be told their child's diagnosis over the phone. This has happened with other Batten children in Scotland and clear guidance must be in place for clinicians.

3 Education

At nursery Reece was well supported. When he started at school, he had high levels of need that could only be met with 1:1 support. Donna wanted him to be active but felt school was risk averse, he was already regularly falling over due to progressive sight loss and Batten. The solution was to confine him to a wheelchair which caused him huge distress. Donna withdrew him from school and had to fight for school staff to visit him at home and provide education support, they came once a week. Children with complex rare diseases sometimes need high levels of support, if schools cannot meet the needs, then alternative education support must be in place.

4 Child Plan meetings (GIRFEC)

These were held every 3-4 months. Donna found these highly distressing as the knowledge around his Batten was not robust and the support for her was sparse. Reece was not attending school, and Donna was his sole carer. 'I stopped attending child plan meetings as they affected my mental health so badly, so the advocacy service and BDFA stepped in for more than 5 years'.
Where a rare and life shortening disease is diagnosed professionals need to be proactive in researching and reaching out to other professionals and charitable organisations for support.

5 Anticipatory Care Planning

As Reece's condition progressed, Donna wanted clear information, advice and support to help her understand how Batten was affecting him and how it would change, to help her plan for Reece's inevitable early death. Donna said 'Drs must factor that Parents are continuously processing the horrendous news of the disease, they must go at our pace and be prepared to answer difficult questions. Rachel House, Chas at home and the BDFA were amazing, without them I felt I had no one on Reece's side'. ACPs must have robust information regarding the growing level of health support, they need to reflect the complex decision-making processes parents go through and they need to be updated in a timely fashion. 'The NHS ACP was very brief it didn't add any value to Reece's journey'.

6 End of Life Support

Donna said 'To make informed choices around appropriate levels of treatment and interventions you need to know the stark reality so you can make the best decisions for your child. I found out when he was already very ill that the children's palliative service out of hours in Highlands and Islands was non-existent. I felt the charities were all proactively supporting me, whereas I saw the NHS /Social care side to be inert and offering only hospital admissions and the children's ward for support out of hours. In the end Reece showed us the way. During his final stay at Rachel House, he became too ill to come home'.

7 Bereavement support

Grief doesn't have a time frame, it changes how it affects people, it needs energy to process and that energy isn't easy to find. Bereavement starts before a child has died and Donna felt that having practical advice and being able to talk about things that would change for her before Reece died would have helped her afterwards, things like planning for his funeral, understanding changes to benefits, Motability vehicles, and the need to go back to employment. She said, 'Nobody wanted to talk about these things in case it upset me, but if someone had started the conversation it would have helped'. Donna feels supported by the BDFA but also believes that bereavement support should be available to all parents and families in their local area, especially as resources seem concentrated along the central belt in Scotland.

Reece's journey highlights practical changes that could make care more compassionate, coordinated and responsive — from diagnosis through bereavement.

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THE BDFA CHECKLIST

Listen to parents • Diagnose earlier • Communicate compassionately
Support inclusion in education • Plan ahead • Provide accessible palliative care
• Offer local bereavement support • Work with families and specialist charities