Coping with complex epilepsy whilst striving for a quality of life for the whole family - facilitating a parent's perspective

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For families receiving an initial diagnosis of epilepsy the focus is all about stopping the seizures. Parents take their child to the doctor and expect them to be able to 'cure' the seizures by giving them medicine. However, in the UK 36% of epilepsy patients have inadequate control of seizures with AEDs[1]. These families for whom seizure freedom is not achieved by medication endeavour to cope with complex drug resistant epilepsy (DRE). For them the seizures become 'just' seizures and it is everything else that surrounds them that takes over day to day living. 'Normal' life has become a thing of the past and it is finding a quality of life (QoL) between seizures that is crucially important for the continued survival of these families.

We also know QoL is an important measure of treatment efficacy, yet there are no validated assessment tools available to take into consideration the subtle changes in QoL in children with DRE that may be considered by their families as huge improvements but cannot be quantified. A recent survey was developed to attempt to better understand exactly what QoL looks like to these parents and in particular how the side effects of management options impact this.

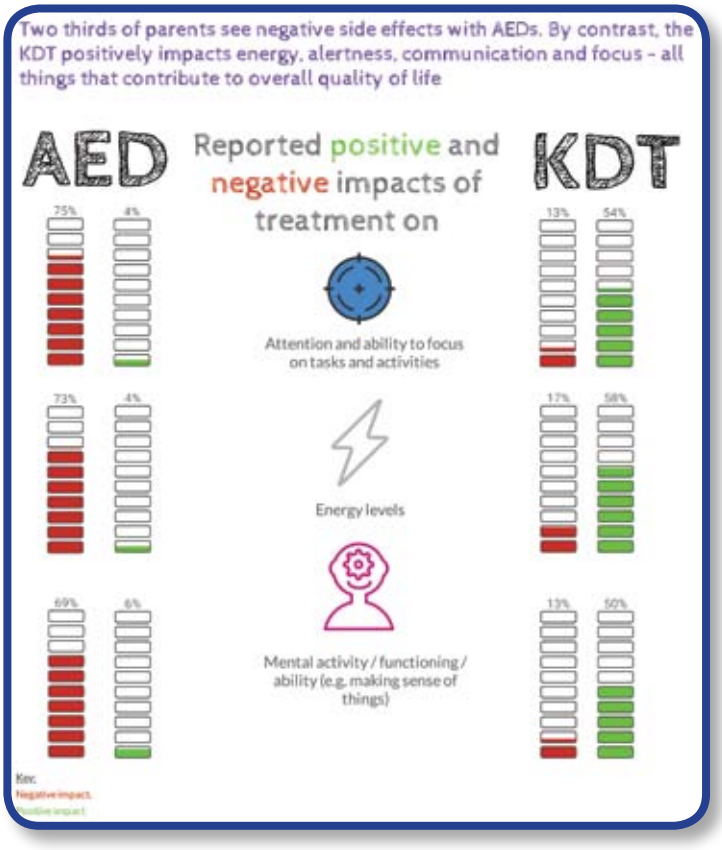
The survey was scripted with 37 questions of differing types including open, closed and free text. The questions were rigorously reviewed by an expert panel of specialist healthcare professionals and families with children with epilepsy to check for sensitivity and content. The survey was then distributed via charity contacts and social media.

51 responses were received from parents of children with epilepsy, 37 of whom experienced the ketogenic diet therapy (KDT) as a management option. The responses were very detailed with over half of parents leaving contact details (unusual for so called anonymous market research which clearly demonstrates the need of these vulnerable families for support and for their voices to be heard).

Key observations from the responses[2]:

42% of responders rated their child's QoL as poor or very poor when related to seizure reduction, happiness and enjoyment, ability to have social interactions with peers and achieve a normal everyday life. The language used was not aspirational and to these families QoL was more so around being normal and being able to do all the things that everyone else does.

Only a quarter of responders reported an improvement in QoL with the introduction of anti-epileptic drugs (AEDs), with 82% reporting negative side effects which impact the whole family.



Over a half (59%) reported that doctors did not recognise the full extent of AED side effects. One family even reported that these side effects were sometimes worse than the seizures themselves.

Side effects related to AEDs, including poor attention span, low energy levels, poor memory and sleepiness were reported in over two thirds of the responders.

49% of parents with children on KDT reported that KDT had improved their child's QoL. Over half reported an improvement in those same measures during dietary management; attention, energy levels and memory, with less anxiety, sadness and depression. Parents also commented that they welcomed the opportunity of KDT to do something for/take part in the care of their child. They felt it was empowering at a time when they felt helpless and unable to do anything to make their child's life happier/better.

CONCLUSION:

Epilepsy continues to be considered a taboo illness and QoL is poor in almost half of responders to this survey. The responses indicate that AEDs impact on QoL more than other therapies like the ketogenic diet which has a perception (rightly or wrongly) as 'difficult'. Despite the challenging nature of the initial introduction of KDT, it is linked to an improvement in QoL with parent reported outcome measures which are not currently measured as part of management efficacy. These families need a QoL in between seizures and so does the child. However, if, when attempting to control the seizures, the extent to which AED side effects impact the child and family could be more fully recognised, and the HCPs and families could find a management plan where improving QoL is the aim, balancing seizure control with AED side effects then Social Service involvement and support would be eased. This is because families would be in a better position to cope and therefore savings would be generated.

The finding from this survey demonstrates that KDT can have a positive impact on QoL over and above seizure reduction and therefore, it begs the question – why is it that the KDT is not considered or offered as a credible epilepsy management plan at an earlier stage?

Furthermore, despite the NICE Guideline[3] recommending referral to a tertiary centre for consideration of other management such as KDT, VNS or surgery after failure of 2 or more appropriately prescribed drugs, over 60% of the children in this survey group had been prescribed 5 or more AEDs. This finding also contradicts the recently updated global consensus statement from the world's leading experts in KDT, which state that if the first 2 medications fail then the diet should at least be discussed with the family. This suggests there is a treatment gap whereby some children are being denied or having delayed access to options that can be as effective as drugs or could indeed be curative such as surgery. This is perhaps corroborated by a third of these families reporting that they had researched and initiated conversations about KDT with their doctor. Research shows that at least 10% of children that go on the diet WILL become seizure free for the rest of their lives, with two thirds of children going on the diet having at least a drop of 50% in their seizures. For a child that is having hundreds of seizures a week, a 50% drop is massive and means there is more of a chance of a life between seizures.

Complex epilepsy is by definition very complicated and requires expert management. For ease of identification of those patients to refer without delay to a tertiary centre for consideration of KDT, the recently updated International Ketogenic Diet Study Group consensus guideline[4] lists the types of syndromes that are known to respond well to KDT. They are listed in the following 2 tables:

Table 1

Epilepsy syndromes and conditions (listed alphabetically) for which KDT has been consistently reported as more beneficial (>70%) than the average 50% KDT response (defined as >50% seizure reduction).

- Angelman syndrome [35,36]
- Complex 1 mitochondrial disorders [30,34]
- Dravet syndrome [14,15]
- Epilepsy with myoclonic-atonic seizures (Doose syndrome) [13,16,17]
- Glucose transporter protein 1 (Glut-1) deficiency syndrome (Glut 1 DS) [7,9-12]
- Febrile infection-related epilepsy syndrome (FIRES) [23-26]
- Formula-fed (solely) children or infants [27,28]
- Infantile spasms [10,39,40]
- Ohtahara syndrome [50-52]
- Pyruvate dehydrogenase deficiency (PDHD) [8]
- Super-refractory status epilepticus [23,25,32,33]
- Tuberous sclerosis complex [20-22]

Table 2

Several conditions (listed alphabetically) in which KDT has been reported as moderately beneficial (not better than the average dietary therapy response, or in limited single-centre case reports).

- Adenylosuccinate lyase deficiency [43]
- CDKL5 encephalopathy [46]
- Childhood absence epilepsy [48]
- Cortical malformations [50,51]
- Epilepsy of infancy with migrating focal seizures [47]
- Epileptic encephalopathy with continuous spike-and-wave during sleep [49]
- Glycogenosis type V [44]
- Juvenile myoclonic epilepsy [45]
- Lafora body disease [37]
- Landau-Kleffner syndrome [40]
- Lennox-Gastaut syndrome [6]
- Phosphofructokinase deficiency [42]
- Rett syndrome [38,39]
- Sabacute sclerosing panencephalitis (SSPE) [41]

Although this is a small survey with some limitations, as the target population surveyed could have an inherent bias for non-pharmacological management, it does serve to highlight the importance of using parent reported outcome measures in research and clinical practice to evaluate treatment efficacy. It further demonstrates that KDT can have a positive impact on QoL over and above seizure reduction and should therefore be considered earlier. With such potential benefits the KDT should no longer be considered as a last resort and should be spoken about in a more positive and proactive way.

DISCLAIMER This survey was undertaken by Nutricia Ltd, a manufacturer of Foods for Special Medical Purposes (FSMPs), to gain a deeper understanding of the challenges faced by families coping with a child with complex epilepsy and therefore not for commercial gain. The findings detailed in this article will facilitate a second more targeted survey in due course.

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