



Robyn

Wes

# Ketogenic Diet in Children with Down Syndrome and Refractory Epilepsy

Joanna Garcia Pierce MD, Tiffani L. McDonough MD, Robyn Blackford RDN, Wesley Lowman RDN, Priyamvada Tatachar MD MBBS

## Introduction

- Down Syndrome (DS) is the most common genetic cause of intellectual disability.
- People with DS have high rates of epilepsy with Infantile Spasms (IS) being the most common seizure type occurring in children.
- Onset of seizures are 6-8 months of age.
- For children with Infantile Spasms, the most promising treatments are: ACTH, Vigabatrin, and the Ketogenic Diet (KD).
- Study Aims:
  - Is the Ketogenic Diet a safe treatment option for those with Infantile Spasms and Down Syndrome?
  - Is the KD efficacious for this population?

## Methods

- Retrospective review of a cohort of patients with DS and refractory epilepsy (including IS) on KD
- Time frame: KD initiations ranged from start date of Feb 2008 through Feb 2019
- Number of patients identified: 7 (86% males)
- Patients were followed in KD clinic at intervals of: 1-month post-diet start and at 6-months
- Labs done at those intervals: BHB, CMP, Cholesterol and Triglycerides

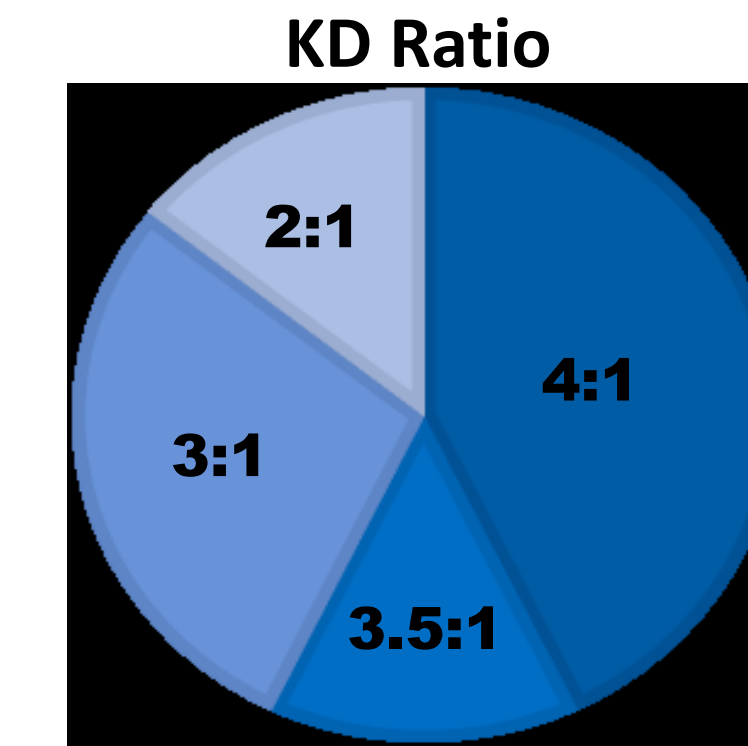
## Results

Patient ID	Age of seizure onset (months) Mean: 10.8m SD: 15.2	Initial daily seizure frequency prior to KD	ASM Number at KD	Age of KD Initiation (months) Mean: 24.4m SD: 27.3	Duration of KD (months) Mean: 20m SD: 13	ASM Number at 6 month follow up	Seizure rate at 6 month follow up
1	5.5	12-50 clusters (8-10 spasms)	2	17	10	2	Decreased to 10 clusters/day
2	7	2-3 clusters (2-5 spasms)	4	15	31	0	Seizure free
3	0	Daily clusters, not quantified	3	18	36	2	Seizure free
4	45	Sporadic	2	86	27	2	Seizure free
5	7	Not quantified	4	12	6	2	Seizure free
6	5	4-5 clusters (2 min each)	3	11	4	N/A	Seizure free at 2m, KD stopped at 4m, Spasms recurred
7	6	1-2 clusters (2-3 spasms)	2	12	12	0	Seizure free

Keywords:

ACTH = adrenocorticotrophic hormone (steroid injection)  
ASM = anti-seizure medication  
BHB = Beta-Hydroxybutyrate (blood ketones)

Seizure Freedom: 5/6



Complications	# of pts/7 (%)
Hyperlipidemia	6 (85.7)
GI Intolerance	3 (42.9)
Acidosis Hypoglycemia	2 (28.6)
Strokes Weight loss Renal Stones	0

The Ketogenic Diet is a high fat and low carbohydrate diet used for the treatment of medically resistant epilepsy.  
KD Ratio → grams Fat : grams Carb + Protein

## Conclusions

- Most achieved seizure freedom while on the KD without any serious complications.
- The KD can be introduced early in patients with DS who have refractory epilepsy, including IS.
- Future research to investigate lipid panel alterations and long-term effects of hyperlipidemia
- Limitations: retrospective review, small study cohort, short follow up time frame

## References

Arya, Kabra. "Epilepsy in Children with Down Syndrome." *Epileptic disorders* 13.1 (2011): 1-7.  
Practice Parameter: Medical Treatment of Infantile Spasms. Report of the American Academy of Neurology and the Child Neurology Society. M. T.Mackay, S. K.Weiss, et al. *Neurology*. May 2004, 62(10) 1668-1681.  
Bough, K.J. and Rho, J.M. (2007), Anticonvulsant Mechanisms of the Ketogenic Diet. *Epilepsia*, 48: 43-58.  
Kwiterovich, Jr PO, Vining EPG, Pyzik P, Skolasky, Jr R, Freeman JM. Effect of a High-Fat Ketogenic Diet on Plasma Levels of Lipids, Lipoproteins, and Apolipoproteins in Children. *JAMA*. 2003;290(7):912-57.

Questions and comments welcome!

Robyn – rblackford@luriechildrens.org    Wes – wlowman@luriechildrens.org