

CASE STUDY 7: ZOE

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Patient Profile

Zoe is a 3 year and 9 month old girl with phenotype consistent with Rett syndrome. She has a lower-middle class family. Her mum studies nursing and her dad is a truck driver.

Background

At the age of 6 months, she began to show visible evidence of global developmental delay together with acquired microcephaly. She presents generalized hypotonia with axial predominance, she achieved cephalic support at 13 months. However, she has not yet achieved autonomous sitting. She had an absence of language until 18 months, when she had started with babbling, a pattern that she later lost. She does not imitate sounds. She has trunk tremor, stereotyped hand movements continually and permanent bruxism during waking hours.

At 2 years and 6 months of age, she began with frequent nocturnal focal seizures and less frequent myoclonic seizures, drug-resistant, medicated with levetiracetam, valproic acid, clobazam and oxcarbazepine.

The electroencephalogram shows diffuse slow waves, multifocal spikes and right lateral spike paroxysms of high-frequency discharge.

A progressive deterioration was evident in the patient since the onset of epilepsy with loss of acquired motor patterns and loss of swallowing requiring nasogastric tube placement. Prior to the onset of epilepsy, she had a normal video swallowing. Left hemi-body focal statuses conditioned an acquired left hemiparesis.

Brain MRI with normal spectroscopy and normal complete spine.

PEAT with normal thresholds.

Normal metabolic study.

She shows normal microarray and normal exome (the exome will be checked).

Clinical Assessment

At 3 years and 3 months (December 2021) in context of refractory status, she begins inpatient ketogenic diet (KD) therapy. Prior to starting ketogenic KD therapy, a video swallowing/swallow was indicated which showed that her swallowing was quite ineffective, therefore, she continued feeding via nasogastric tube. Although prior to her hospitalization she was fed by mouth, with a mixed diet, her mother stated that she accepted all foods in the form of porridge and the feeding time was prolonged. She took a bottle 3 times a day with liquid fluid milk.

She was underweight for her size/height and age.

Dietetic Intervention

At the time of starting therapy:

Her weight was 8.5kg (PC 3) and her length was 83 cm with a BMI of 12.1 kg/m^2

Her weight after hospital discharge 8.37 kg (PC<3)

At the beginning of the treatment she was fed a combination of glucose polymers (polymerase) to complete the carbohydrate and ketocal 4:1 powder requirement. Taking into account the low weight and height/size, a slight increase in the protein intake was calculated. Zoe received 105 kcal/kg/weight. Resulting in a total of 890 kcal/day until she reached an appropriate weight for her size/height and age.

Ratio 1.1 ketocal 4.1 powder 75% of the requirement + polymerase diluted in water and distributed in 6 intakes of 140 cc every 4 hours. Food that tolerates without difficulty, administered by gavage. She maintains normal blood glucose (0.80-0.90 gr/ml) and a slight increase in ketonemia (0.4-0.8 mmol/l) is observed in the afternoon. This indication was maintained for 48 hours.

Ratio 1.5 ketocal 4:1 powder 85% of the requirement + polymerase + fibrum fiber module (dietary fiber 76g; 46g soluble fiber and 30g insoluble fiber) is added by constipation. She maintains normal blood glucose (0.75-0.80 gr/ml) and a slight increase in ketonemia (0.9-1.3 mmol/l) is observed in the afternoon. This indication was maintained for 48 hours.

Ratio 2.1 ketocal 4.1 powder 95% of the requirement + polymer + fibrum blood glucose remains normal (0.70- or.75 gr/ml) increase in ketonemia (1.7-2.3 mmol/l) in the afternoon. Fibrum caused abdominal distension and gas, at least at the beginning, so the same proportion remained. This indication was maintained for 6 days.

Ratio 2.5.1 ketocal 4.1 powder 95% of the requirement + polymerase + fibrum + liquigen (MCT emulsion) blood glucose remains normal (0.70- or 75 gr/ml) increase in ketonemia (2.6-3.2 mmol/l) in the afternoon. This indication was maintained for 4 days.

Ratio 3.1 ketocal 4.1 powder 100% of the requirement + polymerase + fibrum + liquigen the blood glucose remains normal (0.65-0.70 gr/ml) the expected ketonemia (2.8-3.8 mmol/l) was achieved in the afternoon. This is the current indication.

Clinical Outcome

Zoe was discharged from hospital 5 days after starting treatment, with a crisis-free ratio 2.1. We continue to increase the ratio from home by communicating by phone via WhatsApp, with a ketonemia and blood glucose monitor controlled by her mother, who twice a day took a sample telling us the result until we achieved proposed ketonemia.

Weight is checked after 7 days 8.73 kg, after 15 days, 8.95 kg.

After 30 days of starting treatment, the weight was 9.2kg.

After 60 days the weight was 10.1 kg having reached (P3) in weight and height /p age.

Learning Points

After 6 months of KD therapy, Zoe is free from focal seizures, some sporadic myoclonies persist. She improved her general condition, attention, she has less tremors and improved her nutritional status. She is in the process of having a gastrostomy button fitted.

She evacuates normally every 24 or 48 hours without difficulty.

She continues with a ketogenic ratio 3.1 Cal total 1026 to 11.4 kg providing 90 cal/kg/weight.

Current weight 11.4 kg Size 86 cm (P).





Resources

Optimal clinical management of children receiving dietary therapies for epilepsy: Updated recommendations of the International Ketogenic Diet Study Group

Kossoff EH, Zupec-Kania BA, Auvin S, Ballaban-Gil KR, Christina Bergqvist AG, Blackford R, Buchhalter JR, Caraballo RH, Cross JH, Dahlin MG, Donner EJ, Guzel O, Jehle RS, Klepper J, Kang HC, Lambrechts DA, Liu YMC, Nathan JK, Nordli DR Jr, Pfeifer HH, Rho JM, Scheffer IE, Sharma S, Stafstrom CE, Thiele EA, Turner Z, Vaccarezza MM, van der Louw EJTM, Veggiotti P, Wheless JW, Wirrell EC; Charlie Foundation; Matthew's Friends; Practice Committee of the Child Neurology Society. Optimal clinical management of children receiving dietary therapies for epilepsy: Updated recommendations of the International Ketogenic Diet Study Group. Epilepsia Open. 2018 May 21;3(2):175-192. doi: 10.1002/epi4.12225. PMID: 29881797; PMCID: PMC5983110.

Ketogenic diets for drug resistant epilepsy:

Martin-McGill KJ, Bresnahan R, Levy RG, Cooper PN. Ketogenic diets for drug-resistant epilepsy. Cochrane Database Syst Rev. 2020 Jun 24;6(6):CD001903. doi: 10.1002/14651858.CD001903.pub5. PMID: 32588435; PMCID: PMC7387249.

Ketogenic guidelines for infants with refractory epilepsy:

van der Louw E, van den Hurk D, Neal E, Leiendecker B, Fitzsimmon G, Dority L, Thompson L, Marchió M, Dudzińska M, Dressler A, Klepper J, Auvin S, Cross JH. Ketogenic diet guidelines for infants with refractory epilepsy. Eur J Paediatr Neurol. 2016 Nov;20(6):798-809. doi: 10.1016/j.ejpn.2016.07.009. Epub 2016 Jul 17. PMID: 27470655.

International recommendations for the Management of Adults Treated with Ketogenic Diet Therapies:

Cervenka MC, Wood S, Bagary M, Balabanov A, Bercovici E, Brown MG, Devinsky O, Di Lorenzo C, Doherty CP, Felton E, Healy LA, Klein P, Kverneland M, Lambrechts D, Langer J, Nathan J, Munn J, Nguyen P, Phillips M, Roehl K, Tanner A, Williams C, Zupec-Kania B. International Recommendations for the Management of Adults Treated With Ketogenic Diet Therapies. Neurol Clin Pract. 2021 Oct;11(5):385-397. doi: 10.1212/CPJ.0000000000001007. PMID: 34840865; PMCID: PMC8610544.

For the practical aspects of calculating KD, the following books are useful;

Neal E. The classical ketogenic diet. In: Dietary Treatment of Epilepsy Practical Implementation of Ketogenic Therapies, UK: John Wiley & Sons Ltd, 2012, p.70

Kossoff EH, Turner Z, Mackenzie C et al. Calculating the ketogenic diet. In: Ketogenic Diet Therapies for Epilepsy and Other Conditions, 7th edn. New York: Demos Health, 2020