Coexistence of medium chain acyl-CoA dehydrogenase deficiency (MCADD) and Type 1 diabetes (T1D): A management challenge

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Introduction

- Medium chain acyl-CoA dehydrogenase deficiency (MCADD) is an autosomal recessive fatty acid β-oxidation defect.
- The enzyme is important in the breakdown of medium chain fats into acetyl-CoA to produce ketones, alternative energy source when glucose hepatic glycogen stores become depleted during prolonged fasting.
- In MCADD, during periods of fasting/acute illness, there are insufficient ketones to compensate for the glucose energy deficit, resulting in a hypoketotic hypoglycaemia. The accumulation of toxic fatty acids can lead to encephalopathy and sudden death. Management includes avoiding prolonged periods of starvation, consuming high carbohydrate drinks during periods of illness and in symptomatic patients, reversal of catabolism and sustained anabolism by provision of simple carbohydrates by mouth or intravenously.
- Coexistence of MCADD and T1D is rare and there is no causal association. A key goal of management in T1D is achievement of good glycaemic control to reduce risk of long-term complications. This can in some cases increase the risk of hypoglycaemia which can be catastrophic in the presence of MCADD.

Methodology/Case Report

- 15-year old boy with both T1D & MCADD.
- Diagnosed with MCADD at 16-months of age following an episode of diarrhoea and vomiting. He was managed with a frequent feeding regimen. His emergency regimen (during periods of illness) was high carbohydrate drink (SOS). He remained well until age 12 when he was diagnosed with T1D.

Management

- T1D was managed with multiple daily injection therapy as he refused sensor augmented insulin pump therapy.
  - Pre meal dose based on insulin:carbohydrate ratio.
  - Free snack of 15g in between meals in-order to avoid long period of starvation. Should he want a snack more than 15g, we advised he gave insulin for it.
  - Emergency regimen SOS 20 (which contains 40g carbohydrate) during acute illness.
  - Blood glucose target was initially set at 5-9 mmol/l but this was later reduced to 4-7mmol/l.
- His HBA1c varied between 43mmol/mol (6.1%) and 66mmol/mol (8.1%) see fig 1.
- He has had no moderate or severe hypoglycaemia. Care was shared between the diabetes team and inherited metabolic disease specialists.

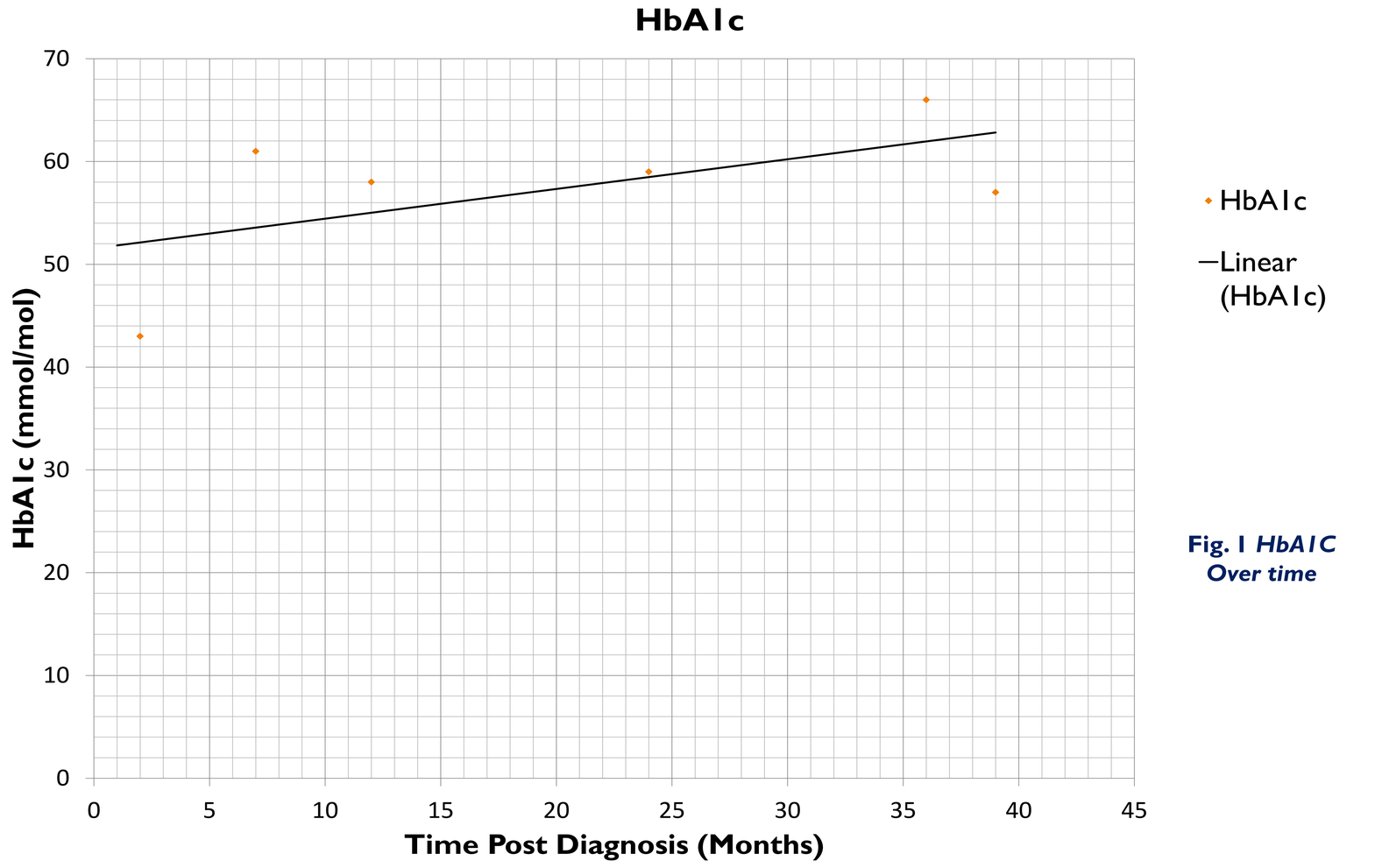

Fig. 1 ***HbA1C Over time***

Conclusion

- Our case describes practical aspects of balancing the concurrent risk of hypoglycaemia whilst trying to achieve good glycaemic control, when T1D and MCADD coexist.
- Shared care between the specialist teams is vital to keeping the patient safe.
- CSII with CGM can help maintain blood glucose levels in range, reduce the frequency of hypoglycaemia and improve treatment satisfaction and quality of life.
- Clinicians can be reassured that as the child gets older, the incidence and frequency of MCADD related hypoglycaemic episodes reduce significantly.

References

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