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THE CHALLENGE OF METABOLIC CONTROL IN CONGENITAL GENERALISED LIPODYSTROPHY TYPE 2 (BERARDINELLI-SEIP SYNDROME)

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INTRODUCTION

Berardinelli-Seip congenital lipodystrophy (BSCL) is a heterogeneous autosomal recessive disorder characterised by an almost total lack of adipose tissue in the body, associated with the progressive development of metabolic complications. There are four different subtypes (I to IV) resulting from mutations in AGPAT2, BSCL2, CAV1 and PTRF genes, respectively. We investigated the characteristics of BSCL2 variants in Sarawak patients.

METHODOLOGY

The clinical features and laboratory indices were obtained by medical interview and medical records review.

RESULTS

Patient 1 was a 19-year-old female diagnosed with lipodystrophy at 5 months, developed dyslipidaemia at 2 years and type 2 diabetes mellitus (T2DM) at 7 years. Her diabetes was difficult to control with metformin and insulin, as evidenced by progressive worsening of HbA1c from 7.4% to 12.1%.

Patient 2 was a 13-year-old male, the younger brother of patient 1. He had dyslipidaemia and T2DM detected at 7 years. Glycaemic control was suboptimal with metformin and insulin, with HbA1c ranging from 9.8% to 14.7%. Subcutaneous premixed human insulin was the initial insulin of choice due to its ease of administration. Inevitably, both progressed to diabetic nephropathy and fatty liver. Recent efforts to intensify glycaemic control with basal-bolus insulin regimen were deemed promising.

Patient 3 was a 7-year-old female who had T2DM at three years. Dyslipidaemia was apparent at one year of age.

Patient 4 was a 3-year-old female, the sibling of patient 3. She had not developed any metabolic complications and was under close surveillance.

Parental consanguinity was identified. All four patients resided in the same village. All of them exhibited a novel homozygous duplication mutation of c.567_573+1dupGAACTCGG.p. in intron 5.

CONCLUSION

The onset of metabolic complications occurs early in the first decade of life in BSCL. Optimal metabolic control is challenging in this condition.