PE-11

USE OF SUBCUTANEOUS LONG-ACTING SOMATOSTATIN ANALOGUE OCTREOTIDE LAR IN A CHILD WITH CONGENITAL HYPERINSULINISM

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INTRODUCTION

Congenital hyperinsulinism (CHI) is characterised by inappropriate insulin secretion and severe hypoglycaemia in infancy. Subcutaneous (SC) short-acting somatostatin analogue is used as second line therapy in diazoxide-unresponsive patients, either as multiple daily injections or via a continuous infusion. Long-acting somatostatin analogues such as octreotide-LAR or lanreotide can be considered after a trial of successful use of short-acting octreotide. Due to the rarity of cases, experiences of its usage in infants and children are mostly limited to small case series. We describe a 7.5 year old child with CHI who was successfully converted from continuous subcutaneous octreotide infusion to intramuscular (IM) octreotide LAR 4-weekly.

RESULTS

Our patient first presented with hypoglycaemia at 2-hour-of-life. He was diazoxide-unresponsive and needed continuous SC octreotide infusion via a pump to maintain normoglycaemia. Genetic testing revealed paternally derived heterozygous ABBC8 non-sense mutation, which suggests a focal form of hyperinsulinism. The family opted to continue medical treatment. In addition, the appropriate imaging (18-F DOPA PET/CT) was not available in the local setting. He had a normal growth rate and neurodevelopment. To improve his quality of life, transition from a continuous subcutaneous infusion of short-acting octreotide (7 mcg/kg/day) to IM octreotide LAR 10 mg every 4 weeks was made at the age of 6 years 11 months. Octreotide infusion was gradually weaned off over 3 weeks with no hypoglycaemia. Frequent home blood glucose pre-meals 4-6 times per day and overnight were in the range of 4-6 mmol/L. The injections were tolerated well with no adverse effects over 6 months. Potential side effects were monitored regularly, which included liver function, thyroid function, IGF-1 and ultrasound abdomen.

CONCLUSION

Long-acting somatostatin analogue should be considered in children with CHI who are diazoxide-unresponsive after a trial of short-acting octreotide. Long term follow-up and monitoring of side-effects are required.

PE-12

CASE REPORT: OVOTESTICULAR DISORDER OF SEXUAL DEVELOPMENT UNMASKED BY ANTIMÜLLERIAN HORMONE

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INTRODUCTION

Disorders of sex development (DSDs) including ovotesticular DSD is a group of rare disorders characterized by abnormalities of chromosomal or discordant gonadal phenotype of internal/external genitalia sexes. Anti-Müllerian hormone (AMH) which is produced exclusively by the prepubertal immature Sertoli cells can be a useful marker for assessment of testicular function in male sex differentiation.

RESULTS

We describe a 1 year 7 month child who presented with atypical genitalia noticed since birth and was investigated for adrenal disorders. The child had neither salt losing crisis nor hypoglycaemia symptoms. The term child was the firstborn of non-consanguineous healthy parents. There was no sexual ambiguity, early neonatal deaths or infertility in the extended family. Physical examination revealed healthy and non-dysmorphic child. The child was normotensive with normal growth and neurodevelopment. External genitalia examination revealed a phallus like structure with a single opening at perineum (Prader stage 4). Only the right gonad was descended. The hCG stimulation test showed partial testosterone response. Serum AMH was above the normal age specific female range indicating the existence of testicular tissue. The karyotype cytogenetic analysis showed the genotype of 46 XX with SRY negative. Ultrasound of gonado-pelvis showed presence of uterus and left inguinal hernia containing left ovary and right inguinal gonad resembling testis. The child was provisionally diagnosed as 46XX ovotesticular DSD.

CONCLUSION

This case revealed the importance of serum AMH as a marker in the evaluation of ovotesticular DSD. The utilization of basal testosterone and gonadotropin stimulation have limited use in the assessment of prepubertal DSD. Serum AMH may provide earlier information in the differential diagnosis of DSDs. Continuous high secretion of AMH by Sertoli cells in a sexually dimorphic pattern during infancy and childhood with high levels in boys and low levels in girls makes AMH such an appealing biomarker of testicular function, henceforth a prior gonadotropin stimulation may not be required in the assessment of DSD cases.