complicated by thyroid storm during the current pregnancy. Her TRAb levels prior to conception were over 23-fold above normal. The infant was born with suppressed cord TSH and markedly elevated TRAb level which is 13-fold above normal. She developed symptoms of neonatal hyperthyroidism in the second week of life and was started on carbimazole and propranolol, which were weaned off by the third week. Subsequent TFTs showed a phase of subclinical hyperthyroidism followed by hypothyroidism by two months of age requiring thyroxine replacement.

CONCLUSION

These cases highlight the diverse presentation of neonatal thyroid dysfunction associated with maternal GD, ranging from transient hypothyroidism to biphasic thyroid disturbances following neonatal hyperthyroidism. High maternal TRAb levels, as seen in Case 2, may serve as a predictor of a more severe case of evolving neonatal thyroid disease. Continuous postnatal monitoring is essential, as thyroid dysfunction may not be evident at birth and can evolve over time. Timely diagnosis and appropriate management are key to prevent complications and supporting optimal neurodevelopmental outcomes.

EP_P026

FAMILIAL DYSALBUMINEMIC HYPERTHYROXINEMIA: A RARE CAUSE OF EUTHYROID HYPERTHYROXINEMIA

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INTRODUCTION

Euthyroid hyperthyroxinemia is a common clinical conundrum. It requires careful assessment to establish an accurate diagnosis. Differential diagnosis of euthyroid hyperthyroxinemia include assay interference, thyroid hormone resistance syndrome, familial dysalbuminemic hyperthyroxinemia (FDH) and TSH-oma.

CASE

A 7-month-old male was referred for incidental finding of persistent euthyroid hyperthyroxinemia. His birth history was unremarkable. Antenatally, his mother did not have any thyroid disorder. His paternal grandmother has been undergoing treatment for hyperthyroidism. Thyroid stimulating hormone (TSH) was elevated at 16.89 mIU/L. Routine prolonged jaundice investigations revealed free thyroxine (FT4) of 33.7 pmol/L and TSH of 7.4 mIU/L. Other investigations were normal. Clinically, he was euthyroid, not dysmorphic, no goitre and thriving well with normal developmental milestones. Repeated thyroid function test (TFT) via standard immunoassay at 2, 3 and 5 months of age showed similar results of high FT4 with unsuppressed TSH. FT3 was not available. TFT using a different assay was not done. Thyroid antibody screening was normal. He was initially suspected of having thyroid hormone resistance syndrome.

Family screening showed similar TFT pattern for his father and sister who were clinically euthyroid. His mother's TFT was normal. His family was referred for confirmatory genetic testing. Whole exome sequencing (WES) for his father identified a pathogenic missense mutation in albumin gene, resulting in the replacement of an arginine with a histidine (p.Arg242His) that is associated with FDH. No genetic testing was done for the children.

CONCLUSION

FDH is a rare cause of euthyroid hyperthyroxinemia. It is an autosomal dominant disorder characterized by an abnormally increased affinity of a mutant albumin molecule to serum thyroxine causing elevated total thyroxine (T4) and elevated or normal FT4 with normal TSH level. Genetic analysis is important to establish diagnosis, to avoid further unnecessary laboratory testing and even inappropriate treatment in FDH.

EP_P027

ATYPICAL GENITALIA IN SILVER-RUSSELL SYNDROME

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INTRODUCTION

Silver-Russell Syndrome (SRS) is a clinically heterogeneous disorder which is often associated with growth restriction.