PP-45

A Case of Paraganglioma and Cyanotic Congenital Heart Disease: A Rare Co-occurrence

https://doi.org/10.15605/jafes.034.S57

Malathi K, Norasyikin AW, Norlela S, Nor Azmi K
Pusat Perubatan Universiti Kebangsaan Malaysia

INTRODUCTION

Pheochromocytoma and paraganglioma (PPGL) are rare neuroendocrine tumors. Cyanotic Congenital heart Disease (CCHD) refers to a collective of heart defects presenting at birth with low level of oxygen in the blood leading to chronic hypoxemia. An association between these two rare diseases has been reported in several case studies.

CASE

A 23-year-old female presented with a history of post-bidirectional cardiopulmonary shunt at the age of 6 for dextrocardia, atrioventricular canal defect and pulmonary stenosis. She had further complications of chronic hypoxemia, secondary polycythemia, cavernous sinus thrombosis and type 2 diabetes mellitus. During follow-up, the patient was found to present with persistent elevation of blood pressure with the triad of palpitations, headaches and diaphoresis. Catecholamine hypersecretion was suspected. Twenty-four hour urine catecholamines revealed norepinephrine 548.2 µg/day [normal value (NV) 12.1 to 85.5], epinephrine 11.6 µg/day (NV 1.7 to 22.4) and dopamine 233 µg/day (NV <496.1). She had no hypertensive retinopathy, but had proteinuria with an estimated glomerular filtration rate of 60 mL/min/1.73 m^2. Thyroid function tests were normal. Other tests showed elevated haemoglobin (20.8 g/dL), low pO2 (51.2 mmHg) and an Hba1c of 7%. Abdominal computerised tomography showed a left large oval paraaortic mass consistent with a paraganglioma with no features suggesting metastasis. After extensive discussion, due to the high-risk procedure and financial limitations, surgery was rejected and the patient family opted for medical treatment. She was treated with rivaroxaban 20 mg OD, prazosin 2 mg TDS, metoprolol 100 mg BD, atorvastatin 40 mg OD, aspirin 100 mg OD and sitagliptin + metformin 50 mg/500 mg OD. Her blood pressure is stable.

CONCLUSION

CCHD patients are at higher risk to develop PPGL due to chronic hypoxia that increases angiogenic factors leading to tumour development. Therefore, active screening and early treatment for PPGL by biochemical or radiological methods may be beneficial for CCHD patients. Clinicians should continue a long-term follow-up to monitor PPGL recurrence if hypoxia is not corrected. Further research is needed for better understanding and revealing the deeper pathogenic connection between hypoxia and PPGL.

PP-46

Misdiagnosis in Discordant Free T4 and TSH Concentrations: Detecting Assay Interference by Method Comparison

https://doi.org/10.15605/jafes.034.S58

Thilakavathiny M, Wan Juani WS
1Hospital Banting, Selangor, Malaysia
2Hospital Putrajaya, Putrajaya, Malaysia

INTRODUCTION

Thyroid function tests (TFTs) are important tools in diagnosing thyroid disorders. In rare cases, TFTs can be misleading due to assay interference which may result in false diagnosis. We describe a patient who presented with neck swelling and falsely elevated free thyroxine (FT4) due to assay interference and discuss a useful strategy to demonstrate assay interference.

CASE

A 40-year-old lady with thyrotoxicosis was previously treated with carbimazole until 2017. Her care was subsequently transferred to another centre. Here, she was found to have elevated FT4 (52.5 pmol/L) and non-suppressed TSH (0.95 mIU/L) using Siemens® platform. She was started on carbimazole 5 mg OD. After a month of treatment, the tests revealed elevated FT4 [127.9 pmol/L, normal value (NV) 11.5 to 22.7] and elevated TSH [34.76 mIU/L, NV 0.55 to 4.78). Carbimazole was increased to 20 mg OD. Her serial FT4 and TSH levels were persistently elevated despite her being euthyroid. In view of discordant biochemical and clinical presentation, thyroid hormone assay interference was suspected. Her TFTs were repeated in 3 different platforms, which revealed low FT4 and elevated TSH concentrations suggestive of severe hypothyroidism.

Assay interferences are usually due to interfering antibodies. By methods of comparison using 3 different two-step immunoassays (Abbott Architect, Beckman DxI 800 and Cobas Roche), all showed low FT4 concentrations and above reference range TSH levels, suggestive of hypothyroidism. Only the Siemens® platform showed a falsely elevated fT4, demonstrating an assay interference with this immunoassay.

CONCLUSION

Assay interference should be considered when there is a discrepancy between clinical picture and biochemical results to prevent inappropriate management. Method of comparison between immunoassays is a useful strategy to demonstrate the presence of autoantibodies as the source of assay interference.