# PP-53

## A FAMILY WITH HEREDITARY PARAGANGLIOMA SECONDARY TO SDHD MUTATION

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### INTRODUCTION

A third of pheochromocytoma and paraganglioma (PPGL) tumours are part of a hereditary syndrome. Hereditary PPGL shows autosomal dominant inheritance with variable penetrance. Genetic testing is recommended in all patients diagnosed with PPGL regardless of age at presentation and family history. SDHD is the most frequently mutated gene in head and neck PGLs and associated tumours have low malignancy rate. This gene is maternally imprinted with silencing of the maternal allele, thus, the risk of developing paragangliomas is limited to offsprings who inherit the pathogenic variant from their father. Paternally inherited pathogenic variants are highly penetrant by age 50. We describe a family with hereditary paraganglioma due to mutation in the SDHD gene.

### RESULTS

The index patient is a 23 year-old female who was diagnosed to have bilateral adrenal pheochromocytoma with carotid and cardiac paragangliomas. Genetic screening revealed that she has SDHD mutation. Further family history revealed that 2 out of 3 paternal aunts have carotid paragangliomas. Her father, youngest of 5 siblings, paternal uncle and grandfather passed away at age 42, 40, 39 years old, respectively, due to severe headache and possible haemorrhagic stroke. Six other family members underwent genetic screening as well. Five family members were positive for SDHD mutation. These include the index patient's elder sister, 3 paternal aunts (2 with carotid paragangliomas) and one male cousin.

### CONCLUSION

Genetic testing for family members of patients with hereditary paragangliomas is recommended after thorough genetic counselling. Genetic testing for first- and second-degree relatives is recommended for SDHD-related paraganglioma. Healthy asymptomatic carriers for the gene mutation should undergo clinical assessment, plasma and/or urine metanephrines and normetanephrines and a combination of whole-body MRI (head and neck, abdominal and pelvic) and PET-CT imaging at initial screening. Thereafter, recommendations for long-term surveillance include annual clinical and biochemical evaluation along with whole body MRI repeated every 2-3 years.

# PP-54

## SERUM ADIPONECTIN AND OTHER PREDICTORS OF NEED FOR INSULIN THERAPY IN GESTATIONAL DIABETES MELLITUS: A PILOT STUDY

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### INTRODUCTION

The prevalence of gestational diabetes mellitus (GDM) is increasing in Malaysia. Adiponectin is an adipokine that is expressed in adipose tissues and placenta. Plasma adiponectin levels are decreased in several metabolic disorders, including obesity, inflammatory states, insulin resistance, and type 2 diabetes. To our knowledge, there are no published reports on the association between plasma adiponectin levels and need for insulin therapy in GDM. The aim of this study was to assess the association of 1) adiponectin and 2) other predictors such as BMI and HbA1c; with the need for insulin therapy in GDM.

### METHODOLOGY

In this prospective pilot study, we recruited women with GDM from combined antenatal clinic. Demographic, anthropometric and clinical data were obtained during the interview. Blood was drawn for insulin, c-peptide, adiponectin and triglyceride at recruitment.

### RESULTS

Of the 142 women included in this study, 16.2% required insulin therapy and 83.8% of patients were able to maintain adequate glycaemic control with diet. We did not find adiponectin at GDM diagnosis to be a significant predictor of need for insulin therapy in both univariate and multivariate analyses. The most robust significant correlation of adiponectin in mothers with GDM ($r > 0.5$) was an inverse association with HOMA IR and fasting insulin which is reflective of insulin resistance. Significant associations of insulin requirement in univariate analysis included history of GDM, history of insulin-requiring GDM and glycaemic variables at diagnosis (higher fasting, 2-hour glucose, AUC glucose). Upon multivariate analysis after adjusting for pre-pregnancy BMI and maternal insulin resistance, only Chinese ethnicity (OR= 4.17, CI 1.32-13.16), history of GDM requiring insulin therapy (OR 10.67, CI 1.78-63.90), and AUC glucose (OR=2.14, CI 1.32-3.45) were significantly associated with increased need for insulin therapy.