

Progress report for Dr Lucia Gagliardi
Endocrine Society of Australia Higher Research Degree Scholarship recipient: January
2008-August 2008

My PhD program comprises three principal studies.

Vasopressin-sensitive ACTH-Independent Macronodular Adrenal Hyperplasia: Clinical and Genetic Studies of Three Kindreds

Phenotyping

This is now complete for the first two families, and is underway for the third family.

Adrenal tumour studies

I have performed qRT-PCR of the vasopressin receptors on the adrenal tumours for the first family.

Microarray studies of tumour gene expression are complete – I am currently analysing the data to determine if there are particular genes or pathways that are up- or down-regulated in the tumours, and am comparing this to the currently published literature.

Manuscript submission

I have submitted a manuscript detailing the phenotyping data for the three families and adrenal tumour molecular studies – the manuscript is currently under revision.

Genetic studies

Leucocyte DNA collection has been completed and in the next few weeks I will send this for SNP typing.

Presentations relating to this study:

Endocrine Society's Annual Meeting, San Francisco, 2008

Poster Presentation – Vasopressin-sensitive ACTH-Independent Macronodular Adrenal Hyperplasia: Clinical Studies of a Large Australian Kindred

Endocrine Society of Australia Annual Scientific Meeting, Melbourne, 2008

Oral Presentation - ACTH-Independent Macronodular Adrenal Hyperplasia: A Familial Disease? Detection of Early Manifestations

Screening for Cushing's Syndrome and Primary Aldosteronism in Type 2 Diabetes Mellitus

Recruitment

I have recruited 70 of the 100 patients needed for this study.

Continuous Subcutaneous Hydrocortisone Infusion Therapy in Addison's Disease

Preliminary studies

I have performed the dose-response evaluation studies to ascertain endogenous cortisol increases after various bolus doses of hydrocortisone.

Materials

All materials for this study have been sourced, and all logistical issues resolved.

Recruitment

I am currently approaching consultants for possible suitable candidates for this study.