Author’s response to reviews

Title: A putative role for the aryl hydrocarbon receptor (AHR) gene in a patient with cyclical Cushing’s disease

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Author’s response to reviews:

Ciarán Fitzpatrick, BSc MSc PhD
Editor
BMC Endocrine Disorders

Dear Professor Fitzpatrick,

Re: ‘A putative role for the aryl hydrocarbon receptor (AHR) gene in a patient with cyclical Cushing’s disease’

Thank you for considering the above manuscript for publication in BMC Endocrine Disorders. This case study proposes a novel role for the aryl hydrocarbon (AHR) gene in cyclical Cushing’s disease. We report a rare, damaging germline AHR variant in a man with cyclical Cushing’s disease and present evidence for pathogenicity of this variant and the role of AHR in both the clock system and pituitary tumorigenesis. This is a preliminary but interesting and important advance as the molecular basis of cyclical Cushing’s syndrome is neither known, nor has it been studied, apart from PRKARA1A mutations in a subset related to primary pigmented nodular adrenocortical disease. This paper also represents the first description of an AHR mutation in the setting of pituitary adenoma.

Please note that after editorial feedback at our initial submission, we have revised the manuscript to meet the Case Report format of BMC Endocrine Disorders. We believe this case is highly instructive as our findings shed new light on the possible pathogenesis of cyclical Cushing’s. As before, we believe the manuscript will be of interest to your wide readership of both academic and clinical
endocrinologists. We anticipate it being the starting point for further research into the role of AHR in pituitary tumorigenesis and cyclical Cushing’s syndrome.

We, the authors, hereby confirm that we approve the manuscript for submission and have no competing interests. The manuscript has not been published, or submitted for publication elsewhere.

Yours sincerely,

Dr Sunita De Sousa
On behalf of all authors