

ORAL PRESENTATION



Clinical and mutational spectrum of patients with congenital lipoid adrenal hyperplasia in Southeast Asia

Kansuda Ariyawatkul^{1*}, Patra Yeetong¹, Somchit Jaruratanasirikul², Kah Yin Loke³, Pairunyar Nakavachara⁴, Chawkaew Kongkanka⁵, Taninee Sahakitrungruang¹

From 8th APPES Biennial Scientific Meeting Darwin, Australia. 29 October – 1 November 2014

Aims

Mutations in Steroidogenic Acute Regulatory protein (StAR) cause congenital lipoid adrenal hyperplasia (lipoid CAH), characterized by absent steroidogenesis, potentially lethal salt loss, 46,XY sex reversal and massively enlarged adrenals engorged with cholesterol esters. Nonclassic lipoid CAH is a recently recognized disorder caused by StAR mutations that retain partial function. We aim to delineate the clinical and mutational spectrum of StAR mutations in patients with lipoid CAH.

Methods

The entire coding regions of the StAR gene were assessed by polymerase chain reaction and sequencing analysis.

Results

There were 10 patients of lipoid CAH had mutations in the StAR gene with 5 novel mutations (p.P230L>WfsX, IVS6-1G>A, IVS3+(2-3)insT, p.W147R, p.Q264R). Eight patients had classic lipoid CAH presenting with adrenal crisis during early infancy (range of onset 3-11 months of age). Two siblings had nonclassic phenotypes with later onset adrenal insufficiency without disordered sex development. Adrenal enlargement by imaging was demonstrated in only 3 cases of classic lipoid CAH. The functional studies of novel StAR mutations are being under investigation.

Conclusion

StAR mutations may not be rare in Southeast Asian population. There is a broad clinical spectrum of StAR

¹Chulalongkorn University, Bangkok, Thailand

mutations varying from early onset adrenal insufficiency to late onset of glucocorticoid deficiency with only mild defects in mineralocorticoid and sex steroid synthesis. Adrenal gland enlargement is not pathognomonic for lipoid CAH.

Authors' details

¹Chulalongkorn University, Bangkok, Thailand. ²Prince of Songkla University, Songkhla, Thailand. ³KTP- National University Children's Medical Institute, National University Hospital, Singapore. ⁴Siriraj Hospital, Mahidol University, Bangkok, Thailand. ⁵Queen Sirikit National Institute of Child Health, Bangkok, Thailand.

Published: 28 April 2015

doi:10.1186/1687-9856-2015-S1-O55 Cite this article as: Ariyawatkul *et al.*: Clinical and mutational spectrum of patients with congenital lipoid adrenal hyperplasia in Southeast Asia. International Journal of Pediatric Endocrinology 2015 2015(Suppl 1):055.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

) BioMed Central

Submit your manuscript at www.biomedcentral.com/submit



© 2015 Ariyawatkul et al; licensee BioMed Central Ltd. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http://creativecommons.org/licenses/by/4.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/publicdomain/zero/1.0/) applies to the data made available in this article, unless otherwise stated.

Full list of author information is available at the end of the article