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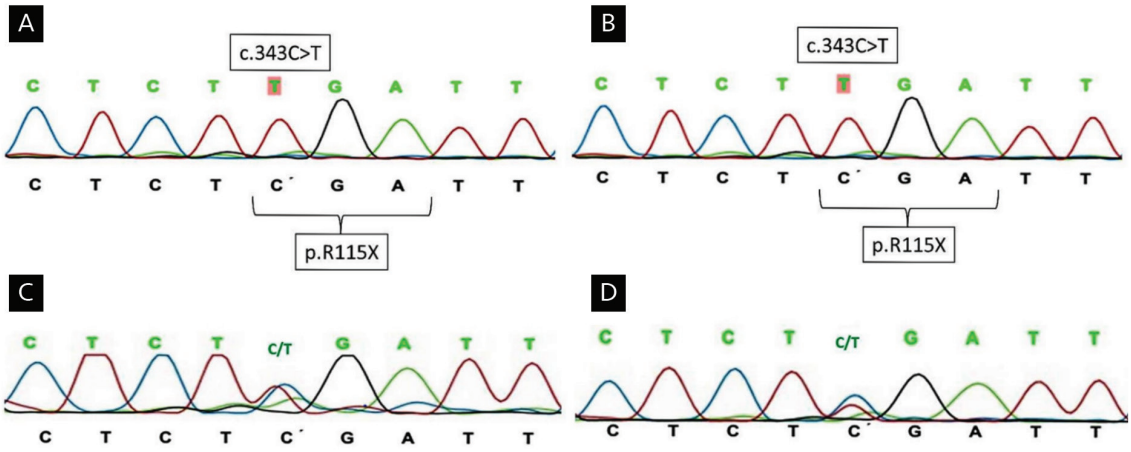

A) (Case 1), B) (Case 2): A novel homozygous nonsense pathogenic variant p.R115X (c.343 C > T) was detected in the CYP19A1 gene sequence analysis. C) (Mother), D) (Father): The parents were heterozygous for the same mutation

Aromatase Deficiency in Two Siblings with 46,XX Karyotype Raised as Different Genders: A Novel Mutation (p.R115X) in the CYP19A1 Gene

Özen S et al.

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damla.goksen@ege.edu.tr ORCID: orcid.org/0000-0001-6108-0591

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korcandemir@gmail.com ORCID: orcid.org/0000-0002-8334-2422

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samim.ozen@ege.edu.tr
ORCID: orcid.org/0000-0001-7037-2713

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serap.turan@marmara.edu.tr ORCID: orcid.org/0000-0002-5172-5402

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Contact
Address: Molla Gürani Mahallesi
Kaçamak Sokak No: 21 34093
Fındıkzade, İstanbul-Turkey
Phone: +90 (212) 621 99 25
Fax: +90 (212) 621 99 27
E-mail: info@galenos.com.tr
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Reviews

Original Articles

Case Reports

CONTENTS

JCRPE Journal of Clinical Research in Pediatric Endocrinology
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CONTENTS