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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