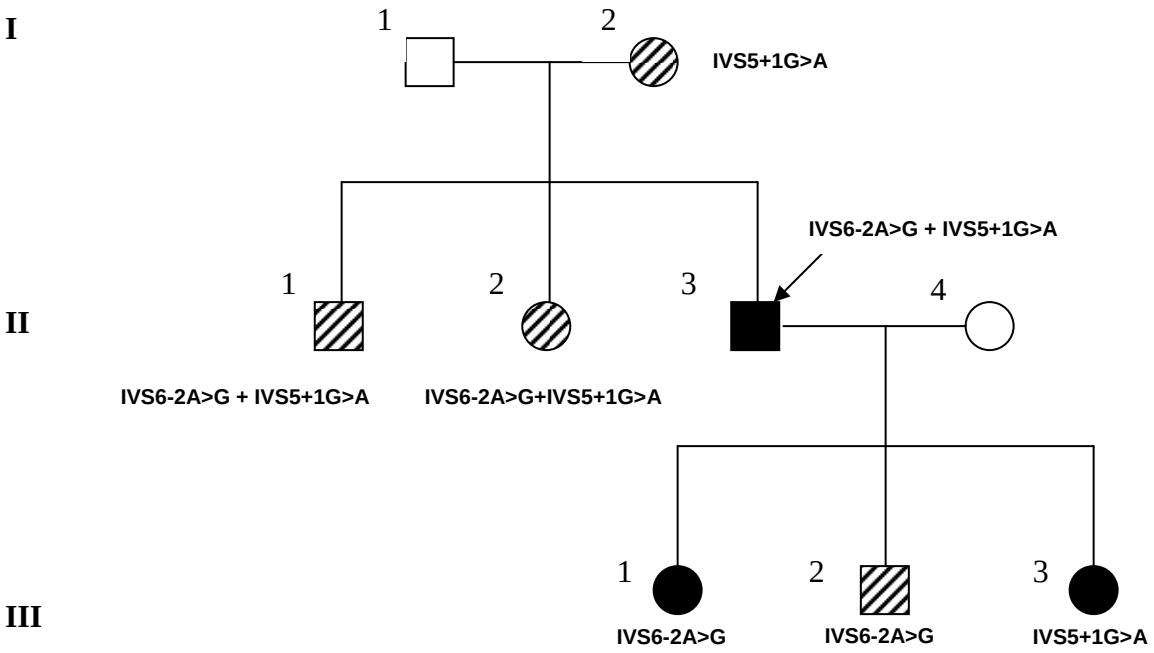
SUPPLEMENTAL DATA

Sequencing of the 12 exons of *CYP24A1* gene was performed using dye termination chemistry (Big Dye Terminator with a model 3730xl DNA sequencer; Applied Biosystems, CA). Primer sets for PCR were designed using a web-based design tool Primer3 (http://frodo.wi.mit.edu) **(Supplemental Table 1)**. Intronic primers covering sequences of interest were designed at least 30 bp away from the intron/exon boundaries. PCR was carried out using AmpliTaq Gold DNA Polymerase (Applied Biosystems) based on the standard protocol. After PCR, the amplicons were treated with the ExoSAP-IT (USB) to degrade unincorporated PCR primers and deoxynucleotide triphosphates. The treated products were mixed with 5 picomoles of the forward or reverse PCR primers for sequencing. DNA sequence variants were identified using Mutation Surveyor Analysis Software (Soft Genetics).

Supplemental Table 1. Primers used to sequence the *CYP24A1* gene

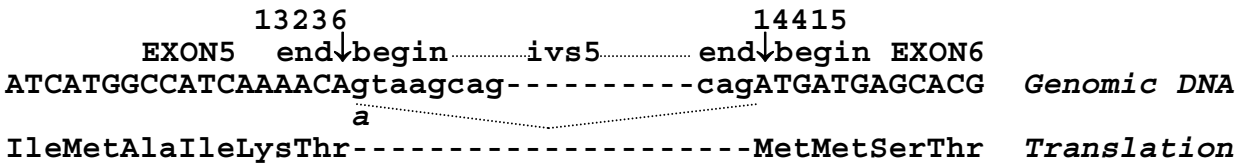
	Forward Primer		Reverse Primer
CYP24A1 Exon1_1_f	AGGGCATGCTCTGTCTCC	CYP24A1 Exon1_1_r	AAGGCAGGAGGATGGGG
CYP24A1 Exon1_2_f	CCCTCTTTGCTTCCTTTTCC	CYP24A1 Exon1_2_r	ATGTCGGGGAGGGTTTG
CYP24A1 Exon2_f	GAGGAAGGAGGCGGGAG	CYP24A1 Exon2_r	CCGTCAGGCTCATCAGGTC
CYP24A1 Exon3_f	GCTGGAGTATTTCTGCATCTCC	CYP24A1 Exon3_r	CCACCAATATCCCTATGTCCC
CYP24A1 Exon4_f	ATGCGATGTAGCAAGACCTG	CYP24A1 Exon4_r	TGCCTGTTTACAAAAGAGTTGTC
CYP24A1 Exon5_f	GGCATAGAATTGAGTCTTTAATAACC	CYP24A1 Exon5_r	TGGGAATCACTGTGAAGTTCTG
CYP24A1 Exon6_f	CCTCTTCCAGAACGAACATTG	CYP24A1 Exon6_r	TGAAGCTCCAGACACGGG
CYP24A1 Exon7_f	TGCAAGAAGGAGTTTGGACTG	CYP24A1 Exon7_r	TGAATCCCAGTGAAATGAATG
CYP24A1 Exon8_f	TTGCAGAATAAGGTGGTGGG	CYP24A1 Exon8_r	TAATTAGCTAGGGGAAGCCG
CYP24A1 Exon9_f	AATCTGCATTCCCATTGACAC	CYP24A1 Exon9_r	CAAAGTCTAGGGAGATCTGGTG
CYP24A1 Exon10-11_f	CAATTTTGCCATTCAAAGGTC	CYP24A1 Exon10-11_r	GCTCATCCCTCGTCATTCTC
CYP24A1 Exon12_1_f	CCGGAAAGCAAACTTCAAAC	CYP24A1 Exon12_1_r	AACAAAATAATGCCCCAGTG
CYP24A1 Exon12_2_f	GCTGGGAGTAATACTGACAATCC	CYP24A1 Exon12_2_r	TATTGCATGCATTTCTGTGC
CYP24A1 Exon12_3_f	TTAGGATCTGTGGTGCAGGG	CYP24A1 Exon12_3_r	TTTGTGATATAGGGCTTGTAGGC

Supplemental Figure 1. Transmission of *CYP24A1* mutations in the affected family. Solid squares or circles indicate a clinical phenotype with biochemical abnormalities; shaded squares or circles represent at biochemical phenotype without reported clinical findings.



Supplemental Figure 2: Splice site mutations observed in affected family (*CYP 24A1*, NG_008334.1)

Splice Site Mutation 1: IVS5+1G>A



Splice Site Mutation 2: IVS6-2A>G