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# Supplemental Data

# Next-Generation Sequencing Reveals Deep Intronic Cryptic *ABCC8* and *HADH* Splicing Founder Mutations Causing Hyperinsulinism by Pseudoexon Activation

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**Table S1. Number of Variants Identified in the 93,929 kb Sequenced around *HADH* from an Individual with Reduced HADH Enzyme Activity after Filtering Steps**

| | Number of Variants Identified in the Proband |
|---|---|
| Total | 123 |
| Exclude variants present in Nov version of 1000 genomes or dbSNP132 | 22 |
| Exclude indels in homopolymer stretches | 5 |
| Exclude variants homozygous in unaffected sibling | 3 |
| Conserved (GERP > 2) or predicted to create cryptic splice site | 1 |

Seventeen putative novel indels were identified, but all occurred in homopolymeric stretches (defined as a run > 6 of the same base) and are likely artefacts caused by long range PCR slippage and/or alignment errors. Cryptic splice sites were assessed using Alamut version 2.1 (Interactive Biosoftware, Rouen, France).

**Table S2. Number of Variants Identified in the 116,738 kb Sequenced around *ABCC8* and *KCNJ11* from Two Individuals after Filtering Steps**

| | Number of Variants from Person 1 with Focal Lesion | Number of Variants from Person 2 with Focal Lesion | Number of Shared Variants |
|---|---|---|---|
| Total | 90 | 225 | 83 |
| Exclude variants present in Nov version of 1000 genomes or dbSNP132 | 9 | 10 | 9 |
| Novel heterozygous variants after excluding indels in homopolymer stretches | 3 | 4 | 1 |
| Conserved (GERP > 2) or predicted to create cryptic splice site | 1 | 1 | 1 |

Cryptic splice sites were assessed using Alamut version 2.1 (Interactive Biosoftware, Rouen, France).