Title: *KCNQ3*-Related Disorders *GeneReview* Table 3

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

**Table 3. Overview of the Available Genetic, Clinical, and Functional Data from Families Carrying *KCNQ3* Mutations: BFNE and BFIE**

| DNA Nucleotide Change | Amino Acid Change | Localization | BFNE | BFIE | Additional Clinical Data | Functional Effects | Reference |
|---|---|---|---|---|---|---|---|
| c.895G>A | p.Glu299Lys | S5-S6 linker region; Pore | + | - | This variant segregates with the BFNE phenotype in 4/5 sibs of a three-generation family; possibly contributes to the Rolandic epilepsy occurring in this family | Reduction in current amplitude | Neubauer et al [2008]; Hahn & Neubauer [2009] |
| c.914A>G | p.Asp305Gly | | + | - | | Reduced current amplitude of heteromeric channels | Singh et al [2003] |
| c.925T>C | p.Trp309Arg | | + | - | | Homomeric channels lacked potassium current and hereromeric channels displayed a dramatic reduction of current | Hirose et al [2000]; Uehara et al [2008]; Sugiura et al [2009] |
| c.929G>T | p.Gly310Val | | + | - | | --- | Charlier et al [1998] |
| c.950T>C | p.Ile317Thr | | + | - | Moderate psychomotor delay | Reduction in current amplitude | Soldovieri et al [2014] |
| c.988C>T | p.Arg330Cys | | + | - | The same variant occurred in two families | --- | Li et al [2006]; Li et al [2008]; Fister et al 2013] |

| DNA Nucleotide Change | Amino Acid Change | Localization | BFNE | BFIE | Additional Clinical Data | Functional Effects | Reference |
|---|---|---|---|---|---|---|---|
| c.1403A>G | p.Asn468Ser | C-terminus | - | + | Three siblings affected | No effect; possibly a non pathogenic variant | Singh et al [2003] |
| c.1720C>T | p.Pro574Ser | | + | - | Found in a typical BFNE family also carrying a *KCNQ2* missense mutation likely pathogenic.<br>Also detected in other complex phenotypes (see Table 2); in fact, it has been detected in four individuals with rolandic epilepsy and in one with rolandic epilepsy and moderate psychomotor delay, in 8/455 patients with idiopathic generalized epilepsies (IGE; absent in 454 controls), and in three patients with autism spectrum disorders, with no additional neurologic features. In the latter study, this variant has been detected at low frequency (0.2-0.4%) also in control individuals. | No difference versus wt; possibly a variant of unknown significance; reduced current amplitude only when co-expressed with Kv7.5 | Neubauer et al [2008]; Hahn & Neubauer, [2009]; Miceli et al [2009]; Lemke et al [2012]; Gilling et al [2013] |
| c.2338C>T | p.Arg780Cys | | - | + | | --- | Zara et al [2013] |
| c.2462A>G | p.Asn821Ser | | + | - | Found in a typical BFNE family also carrying a *KCNQ2* deletion/insertion likely pathogenic; the *KCNQ3* variant does not co-segregate with the disease | No difference versus wt; possibly a variant of unknown significance | Bassi et al [2005] |

BFNE: benign familial neonatal epilepsy

BFIE: benign familial infantile epilepsy

FS: febrile seizures

Reference sequences: NM_004519.3 (KCNQ3), cDNA numbering begins with +1 as the A of ATG initiation codon.

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