CPIC Term Standardization part 2: *MT-RNR1* final termS

After a series of conference calls and surveys, the MT-RNR1 Gene/Disease and PGx expert panels have agreed on terminology for *MT-RNR1* and aminoglycoside-induced hearing loss. Terms that will be used to describe CPIC clinical function are in red font and phenotype terms are in blue font in Table 1 below. The tables below are how these terms will be displayed in the upcoming CPIC *MT-RNR1* guideline. Send feedback on the terms to Kelly Caudle at Kelly.caudle@stjude.org. Please note, CPIC is only seeking feedback on the terminology used to describe allele function and phenotype.

Table 1. Assignment of MT-RNR1 phenotype based on genotype

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| --- | --- | --- |
| **Likely phenotype** | **Genotypes** | **Example genotypes** |
| MT-RNR1 increased risk of aminoglycoside-induced hearing loss  | Individuals with a *MT-RNR1* variant associated with an increased risk of aminoglycoside-induced hearing loss  | m.1095T>Cm.1494C>Tm.1555A>G |
| MT-RNR1 normal risk of aminoglycoside-induced hearing loss  | Individuals with no detectable *MT-RNR1* increased risk variant or a *MT-RNR1* variant associated with normal risk of aminoglycoside-induced hearing loss  | m.827A>G |
| MT-RNR1 uncertain risk of aminoglycoside-induced hearing loss  | Individuals with a *MT-RNR1* variant associated with an uncertain risk of aminoglycoside-induced hearing loss | m.663A>Gm.669T>Cm.747A>Gm.786G>Am.807A>Gm.807A>Cm.839A>Gm.896A>Gm.930A>Gm.951G>Am.960C>delm.961T>Gm.961T>delm.961T>del+Cnm.988G>Am.1189T>Cm.1243T>Cm.1520T>Cm.1537C>Tm.1556C>T |

CPIC MT-RNR1 allele functionality table:

