Title: Tuberous Sclerosis Complex *GeneReview* – Table 1, Details
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Summary of findings in more than 4200 individuals with TSC and their families in whom disease-causing mutations have been identified

- Jones et al [1999] identified exonic and whole-gene deletions in *TSC1* and *TSC2* and small mutations in 120 of 150 (80%) individuals with TSC, of whom 130 represented simplex cases (i.e., individuals who have no family history of TSC) and 20 were familial cases.
- In a study of 38 familial cases, 183 simplex cases, and three of unknown status, Dabora et al [2001] identified small mutations in either *TSC1* or *TSC2* in 166 (74%) probands.
- Using mutation scanning and direct sequencing, Southern blotting, and FISH analysis in 490 families with TSC, Sancak et al [2005] identified small mutations in either *TSC1* or *TSC2* in 342 (70%).
- Using mutation scanning and direct sequencing to screen for mutations in 325 families who met diagnostic criteria for TSC, Au et al [2007] identified 243 (75%) who had small mutations in either *TSC1* or *TSC2*.

References

Au KS, Williams AT, Roach ES, Batchelor L, Sparagana SP, Delgado MR, Wheless JW, Baumgartner JE, Roa BB, Wilson CM, Smith-Knuppel TK, Cheung MY, Whittemore VH, King TM, Northrup H. Genotype/phenotype correlation in 325 individuals referred for a diagnosis of tuberous sclerosis complex in the United States. *Genet Med*. 2007; 9:88-100.

Dabora SL, Jozwiak S, Franz DN, Roberts PS, Nieto A, Chung J, Choy YS, Reeve MP, Thiele E, Egelhoff JC, Kasprzyk-Obara J, Domanska-Pakiela D, Kwiatkowski DJ. Mutational analysis in a cohort of 224 tuberous sclerosis patients indicates increased severity of TSC2, compared with TSC1, disease in multiple organs. *Am J Hum Genet*. 2001; 68:64-80.

Jones AC, Shyamsundar MM, Thomas MW, Maynard J, Idziaszczyk S, Tomkins S, Sampson JR, Cheadle JP. Comprehensive mutation analysis of TSC1 and TSC2-and phenotypic correlations in 150 families with tuberous sclerosis. *Am J Hum Genet*. 1999; 64:1305-15.

Sancak O, Nellist M, Goedbloed M, Elfferich P, Wouters C, Maat-Kievit A, Zonnenberg B, Verhoef S, Halley D, van den Ouweland A. Mutational analysis of the TSC1 and TSC2 genes in a diagnostic setting: genotype--phenotype correlations and comparison of diagnostic DNA techniques in Tuberous Sclerosis Complex. *Eur J Hum Genet*. 2005; 13:731-41.