

[All-cause mortality and survival in adults with 22q11.2 deletion syndrome](#)

Van L, Heung T, Graffi J, Ng E, Malecki S, Van Mil S, Boot E, Corral M, Chow EWC, Hodgkinson KA, Silversides C, Bassett AS. *Genet Med.* 21(10):2328-2335, 2019.

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As information is limited on long term outcomes in 22q, we studied mortality and survival in 309 adults with 22q and their 1014 unaffected parents and siblings. The results showed that the probability of survival to age 45 years was approximately 95% for those with no major congenital heart defect, and 72% for those with a major heart defect. Although the 22q11.2 deletion and more severe forms of congenital heart defects contribute to a significantly lower life expectancy than family-based expectations, a substantial minority of individuals with 22q had outlived both parents. The average age at death was approximately 5 years older than the age we reported 10 years ago for the initial subgroup of 100 patients with 22q.