ACHONDROPLASIA MORTALITY STUDY: A 40 YER FOLLOW-UP STUDY

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Achondroplasia (ACH) is the most common dwarfing condition affecting approximately 1 in 25,000 live births. In 1987, Hecht et al. reported an overall increased mortality with age specific mortality significantly increased up to age 34 years in a large cohort of ACH individuals. Sudden death accounted for an excess of deaths in children less than 4 years and deaths related to cardiovascular disease were increased in adults ages 25 to 54 years. The current study was undertaken as a follow up to this original study. Seven hundred ninety-three ACH individuals from the original study and 75 additional ACH individuals ascertained through the University of Texas Medical School at Houston were included in this study. Collectively, these two cohorts resulted in nearly 20,000 person years of follow-up. The overall mortality was increased in both genders across the 40 years of follow-up with age specific mortality significantly increased up to age 55 years. The average life expectancy was almost 15 years shorter than that of the general population. The mortality and survival in the later 20 years of follow-up for the cohort were not significantly improved compared to the earlier 20 years of follow-up. Sudden death, accidental death, neurological, and cardiac deaths were all significantly increased. The rate of deaths related to cardiovascular disease was over 10 fold higher in ACH individuals ages 25 to 35 than the general population. This study demonstrates that despite advances in the knowledge of the natural history of ACH and health care of this population mortality continues to be increased. Furthermore, the high rates of cardiovascular disease illustrate the need for additional research on the risk factors in the ACH population in order to develop a treatment intervention.