

Facts About AS

How common is Angelman Syndrome?

During the last 20 years, there has been increasing awareness of AS throughout the world. The syndrome is well represented by parent-based support groups in many countries, on individual family websites and on a host of medical and professional information websites. Angelman syndrome has emerged as one of the important syndromes causing neurological impairment and most pediatricians and neurologists now have some awareness of it. How common is Angelman Syndrome. AS has been reported throughout the world among divergent racial groups. In North America, the great majority of known cases seem to be of Caucasian origin. The exact incidence of AS is unknown but the best available data probably come from studies of school age children, ages 6-13 years, living in Sweden, and from Denmark where the diagnosis of AS children in medical clinics was compared to an 8 year period of about 45,000 births. The Swedish study showed an AS prevalence of about 1/12,000 and the Danish study showed a minimum AS prevalence of about 1/10,000. Note that it is desirable to use the term prevalence since estimates of the AS diagnosis have been made in relatively small cohorts of children over various periods of time.

Several reports have tried to address the prevalence of AS among groups of individuals with established developmental delay. The results showed rates of 0%, 1.3% , 1.4% , and 4.8%. The Buckley paper extrapolated their data in order to compare it to the population of the state of Washington (using 1997 Census Bureau figures) and obtained an estimate of 1/20,000, a number similar to that often quoted, but not referenced in terms of methodology, in a 1992 review paper

There appear to be no reported prevalence studies that have screened newborns to detect rates of AS. Population wide prevalence figures would need to take into consideration that longevity in AS is probably reduced (severe mental delay and seizure presence would be risk factors) but no actuarial or other data are available on life span shortening. Likewise, it is not known what percent of individuals with AS are undiagnosed, although this is expected it to be significant. Accordingly, to estimate the number of people with AS living in the society, it would be inaccurate to divide any estimated AS prevalence figure into a total population number.

Given this information, it appears that the prevalence of AS among children and young adults is between 1/10,000 and 1/20,000. It is suggested to use a 1/15,000 figure if a single figure is needed. For population projections, estimates using birth rates can be used. For example, if an area has a birth rate of about 200,000/year it would be estimated that about 13 babies would be born each year with AS.

According to the Clinical Genetic Service, Department of Health, more than 50 cases were diagnosed in Hong Kong. On average three to five new cases were confirmed annually in the last 5 years.