



dsd-LIFE ABSTRACT FOR THE GENERAL PUBLIC

Title Health status in 1040 adults with disorders of sex development (DSD): A European multicenter study

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What did we want to know? As knowledge about health status in adults with disorder of sex development (DSD) is scarce, we wanted to know more about it to improve medical follow-up.

What did we do? We examined persons with DSD and asked them about their health. Due to the rarity of DSD 14 European tertiary centers were involved. 1040 participants with DSD were recruited, of which 301 had Turner syndrome, 224 had Klinefelter syndrome (including 6 46,XX males), 222 had XY-DSD, 21 had XX-DSD (excluding congenital adrenal hyperplasia (CAH) and 46,XX males), 226 had 46,XX-CAH, and 45 had 45,X/46,XY. The health of the persons with DSD was compared to the health of the general European population.

What were the main results? Persons with DSD reported fair to very good general health in 91.4% and only 8.6% reported bad or very bad general health which was slightly worse compared to the general population (94.0% and 6.0%, respectively). Longstanding health issues other than DSD and feeling limited in daily life were reported in 51.0% and 38.6 %, respectively (general population 24.5% and 13.8%). Any disease except DSD was present in 84.3% (general population 24.6%). Men with DSD reported worse health than females with DSD. Persons with Klinefelter syndrome and persons with 46,XX-DSD reported bad general health in 15.7% and 16.7%, respectively, while persons with Turner syndrome and CAH only reported bad general health in 3.2% and 7.4%, respectively. Various other diseases were common in all different DSD conditions but persons with Klinefelter syndrome and Turner syndrome were most affected. However, an early diagnosis of DSD and a healthy lifestyle reduced the risk of other illnesses.

What does that mean? Overall, general health in persons with DSD appeared to be good but a number of medical problems were reported, especially in Klinefelter syndrome and Turner syndrome. Early diagnosis of DSD and a healthy lifestyle seemed to be important and could reduce the risk of other illnesses. Lifelong follow-up at specialized centers is necessary.

Reference:

Falhammar H, Claahsen-van der Grinten H, Reisch N, Slowikowska-Hilczer J, Nordenström A, Roehle R, Bouvattier C, Kreukels BPC, Köhler B; dsd-LIFE group. Health status in 1040 adults with

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