

Turner syndrome with ring x chromosome: do they have a distinct phenotype?

ABSTRACT

In contrast to classic Turner syndrome, Turner patients with ring X chromosome are associated with distinct dysmorphism and are likely to be mentally impaired. Four Turner patients with ring X chromosome were examined for phenotypic features of Turner syndrome and additional dysmorphism. Both patients 1 and 2 are twins with normal intelligence whereas patients 3 and 4 have mental impairment. With the exception of patient 4, the other three patients only have few Turner characteristics. None of the patients have the distinctive dysmorphism previously reported in Turner syndrome with ring X chromosome. Both twins developed spontaneous puberty. Patients 3 and 4 however had no spontaneous puberty. We postulate that this variation may be related to the ring size, the proportion of 45,X and ring X chromosome in cell lines of various body tissues as well as the ability of these rings to be inactivated as a result of lyonisation.

Keyword: Turner syndrome; Ring x chromosome; Mental impairment