Clinical features of girls with turner syndrome in a single centre in Malaysia

ABSTRACT

Objectives. Diagnosis of Turner syndrome in Malaysia is often late. This may be due to a lack of awareness of the wide clinical variability in this condition. In our study, we aim to examine the clinical features of all our Turner patients during the study period and at presentation.

Methodology. This was a cross-sectional study. Thirty-four (34) Turner patients were examined for Turner-specific clinical features. The karyotype, clinical features at presentation, age at diagnosis and physiologic features were retrieved from their medical records.

Results. Patients with 45,X presented at a median age of 1 month old with predominantly lymphoedema and webbed neck. Patients with chromosome mosaicism or structural X abnormalities presented at a median age of 11 years old with a broader clinical spectrum, short stature being the most common presenting clinical feature. Cubitus valgus deformity, nail dysplasia and short 4th/5th metacarpals or metatarsals were common clinical features occurring in 85.3%-94.1% of all Turner patients. Almost all patients aged ≥2 years were short irrespective of karyotype.

Conclusion. Although short stature is a universal finding in Turner patients, it is usually unrecognised till late. Unlike the 45,X karyotype, non-classic Turner syndrome has clinical features which may be subtle and difficult to discern. Our findings underscore the importance of proper serial anthropometric measurements in children. Awareness for the wide spectrum of presenting features and careful examination for Turner specific clinical features is crucial in all short girls to prevent a delay in diagnosis

Keyword: Turner syndrome; Short stature; Webbed neck; Lymphoedema; Karyotype