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Turner syndrome in diverse populations.

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Abstract

Turner syndrome (TS) is a common multiple congenital anomaly **syndrome** resulting from complete or partial absence of the second X chromosome. In this study, we explore the phenotype of TS in **diverse populations** using clinical examination and facial analysis technology. Clinical data from 78 individuals and images from 108 individuals with TS from 19 different countries were analyzed. Individuals were grouped into categories of African descent (African), Asian, Latin American, Caucasian (European descent), and Middle Eastern. The most common phenotype features across all **population** groups were short stature (86%), cubitus valgus (76%), and low posterior hairline 70%. Two facial analysis technology experiments were conducted: TS versus general **population** and TS versus

Noonan **syndrome**. Across all ethnicities, facial analysis was accurate in diagnosing TS from frontal facial images as measured by the area under the curve (AUC). An AUC of 0.903 ($p < .001$) was found for TS versus general **population** controls and 0.925 ($p < .001$) for TS versus individuals with Noonan **syndrome**. In summary, we present consistent clinical findings from global **populations** with TS and additionally demonstrate that facial analysis technology can accurately distinguish TS from the general **population** and Noonan **syndrome**.

KEYWORDS: Turner syndrome; **diverse populations**; facial analysis technology; health disparities

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