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Down syndrome in diverse populations.

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Abstract

Down syndrome is the most common cause of cognitive impairment and presents clinically with universally recognizable signs and symptoms. In this study, we focus on exam findings and digital facial analysis technology in individuals with Down syndrome in diverse populations. Photos and clinical information were collected on 65 individuals from 13 countries, 56.9% were male and the average age was 6.6 years (range 1 month to 26 years; SD = 6.6 years). Subjective findings showed that clinical features were different across ethnicities (Africans, Asians, and Latin Americans), including brachycephaly, ear anomalies, clinodactyly, sandal gap, and abundant neck skin, which were all significantly less frequent in Africans ($P < 0.001$, $P < 0.001$, $P < 0.001$, $P < 0.05$, and $P < 0.05$, respectively). Evaluation using a digital facial analysis technology of a larger diverse cohort of newborns to adults ($n = 129$ cases; $n = 132$ controls) was able to diagnose Down syndrome with a sensitivity of 0.961, specificity of 0.924, and accuracy of 0.943. Only the angles at medial canthus and ala of the nose were common significant findings amongst different ethnicities (Caucasians, Africans, and Asians) when compared to ethnically matched controls. The Asian group had the least number of significant digital facial biometrics at 4, compared to Caucasians at 8 and Africans at 7. In conclusion, this study displays the wide variety of findings across different geographic populations in Down syndrome and demonstrates the accuracy and promise of digital facial analysis technology in the diagnosis of Down syndrome internationally.

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KEYWORDS: diverse populations; down syndrome; facial analysis technology; trisomy 21

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