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Craniofacial Anthropometry and Facial Indices among Nigerian Individuals with Down Syndrome: A Comparative Cross-Sectional Study

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Abstract

Down syndrome is associated with characteristic craniofacial morphology, yet population-specific anthropometric data from Nigeria remain limited. This study compared craniofacial dimensions and facial indices between Nigerian individuals with Down syndrome and apparently healthy controls. A comparative cross-sectional study conducted in Jos, Nigeria, included 30 individuals with Down syndrome and 60 controls aged 4–24 years. Bizygomatic breadth, morphological facial height, and morphological upper facial height were measured using direct anthropometry. Morphological facial and upper facial indices were calculated from these measurements. Group differences were assessed using independent-samples t-tests, with mean differences, 95% confidence intervals, and Cohen's d reported. Bizygomatic breadth was lower among participants with Down syndrome than among controls (118.4 ± 9.73 versus 121.5 ± 8.59 mm), although the difference was not statistically significant ($p = .126$; $d = 0.35$). Morphological facial height (98.67 ± 9.58 versus 103.8 ± 9.02 mm; $p = .015$; $d = 0.55$) and morphological upper facial height (54.87 ± 6.27 versus 59.40 ± 5.12 mm; $p < .001$; $d = 0.82$) were significantly lower in the Down syndrome group. Morphological facial and upper facial indices were also significantly lower ($p = .009$ and $p < .001$, respectively), with the largest effect observed for the morphological upper

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facial index (d = 1.04). Overall, differences were more pronounced in vertical facial dimensions and facial proportions than in transverse facial width. These findings expand population-specific anthropometric evidence and support the usefulness of direct craniofacial anthropometry for characterizing the facial phenotype of Down syndrome among Nigerian individuals.

Keywords: Craniofacial Anthropometry; Down Syndrome; Facial Indices; Facial Morphology; Nigeria

INTRODUCTION

Down syndrome (DS), or trisomy 21, is a chromosomal condition associated with characteristic developmental, medical and morphological features. Craniofacial morphology forms an important component of its recognisable phenotype and reflects alterations in craniofacial growth and development rather than isolated facial anomalies (Bull et al., 2022; Redhead et al., 2023). Recent experimental evidence further suggests that increased dosage of chromosome 21 genes, including DYRK1A, contributes to the altered development of craniofacial structures in DS.

Individuals with DS commonly exhibit distinctive craniofacial characteristics, including midfacial hypoplasia, reduced maxillary dimensions, flattening of the midface and altered facial proportions. Three-dimensional facial analysis has demonstrated reduced dimensions of several facial structures in individuals with DS, while a systematic review and meta-analysis of cephalometric studies reported reduced anterior and posterior facial heights, a shorter and flatter cranial base, and reduced maxillary dimensions compared with unaffected controls (Jayaratne et al., 2017; Vicente et al., 2020).

Anthropometric assessment provides an objective means of quantifying these morphological differences. Measurements such as bizygomatic breadth, morphological facial height and morphological upper facial height describe transverse and vertical facial dimensions, while the morphological facial index and morphological upper facial index express these dimensions proportionally. Such indices are particularly useful because differences in facial proportions may be evident even when individual linear dimensions show substantial overlap between populations. Although three-dimensional stereophotogrammetry offers detailed digital assessment, direct anthropometry remains a practical approach where the study objective is the comparison of defined linear

dimensions and facial proportions. Contemporary Nigerian studies using both direct and three-dimensional methods have also demonstrated appreciable population-, age- and sex-related variation in facial dimensions (Adekunle et al., 2022; Olusanya et al., 2018).

Population-specific assessment is therefore important when characterising craniofacial morphology. Studies among Nigerians without craniofacial abnormalities have shown that facial dimensions and proportions do not necessarily conform to classical norms derived from other populations (Olusanya et al., 2018; Adekunle et al., 2022). Nigerian data specifically relating to DS remain limited. A recent study in Kaduna State identified significant anthropometric differences between children with DS and controls, including reduced morphological facial height; however, the study evaluated a broad range of body and craniofacial measurements rather than focusing specifically on facial width, vertical facial dimensions and their derived proportional indices (Bashir et al., 2026).

This study therefore aimed to compare craniofacial anthropometric measurements and facial indices between Nigerian individuals with Down syndrome and apparently healthy controls. Specifically, the study compared bizygomatic breadth, morphological facial height and morphological upper facial height, as well as the morphological facial index and morphological upper facial index, between the two groups.

MATERIALS AND METHODS

Study Design and Setting

This was a comparative cross-sectional study conducted in Jos, Plateau State, North-Central Nigeria. Participants with Down syndrome were recruited from five schools in Jos: Open Doors Special Education Centre, Otana International School, Model Teaching Centre, Othneal Special School, and Islamiya Primary School, Bauchi Road. Apparently healthy controls were recruited from Islamiya Primary School, Bauchi Road, Jos.

Study Participants and Recruitment

The study comprised 30 individuals with Down syndrome and 60 apparently healthy controls aged 4–24 years. The Down syndrome group consisted of 17 males and 13 females, and the control group of 34 males and 26 females. All available individuals with Down syndrome from the participating schools were recruited (total enumeration), and

apparently healthy controls were recruited from Islamiya Primary School using simple random sampling in an approximate 2:1 control-to-case ratio. The diagnosis of Down syndrome was based on clinical diagnosis. Individuals with a history of craniofacial trauma or surgery, or with congenital anomalies other than Down syndrome likely to affect facial dimensions, were excluded.

Sample Size

The number of participants with Down syndrome was determined by the total number of eligible individuals available at the participating schools during the study period rather than by a formal a priori power calculation; all such individuals were therefore invited to participate. A 2:1 ratio of controls to participants with Down syndrome was adopted to improve the precision of the between-group comparisons.

Anthropometric Measurements and Facial Indices

Craniofacial anthropometric measurements were obtained using standardized direct anthropometric techniques following established craniofacial anthropometric protocols (Farkas, 1994). Participants were examined with the head maintained in the natural head position, and linear measurements were obtained using appropriate anthropometric calipers and recorded in millimeters. Standard anatomical landmarks were identified before each measurement. To improve measurement reliability, each anthropometric measurement was obtained twice by the same investigator, and the average of the two measurements was used for analysis. Intra-observer reliability was assessed from the duplicate measurements using the technical error of measurement (TEM) and coefficient of reliability (R). The coefficient of reliability was 0.97 (relative TEM = 2.1%), indicating excellent measurement consistency

Bizygomatic breadth (zy–zy) was measured as the maximum horizontal distance between the right and left zygion using a spreading caliper.

Morphological facial height (n–gn) was measured as the vertical distance between the nasion and the gnathion using a sliding caliper.

Morphological upper facial height (n–pr) was measured as the vertical distance between the nasion and the prosthion using a sliding caliper.

Measurements were recorded in millimetres.

Anthropometric landmarks

The landmarks used were:

Zygion (zy): the most lateral point of the zygomatic region.

Nasion (n): the midline point at the junction of the frontal and nasal regions.

Gnathion (gn): the lowest midline point on the chin.

Prosthion (pr): the most anterior midline point of the upper alveolar region.

The locations of the landmarks and the corresponding linear measurements are illustrated in Figure 1.

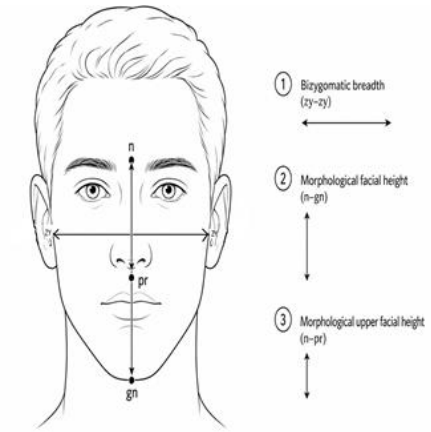


Figure 1. Craniofacial anthropometric landmarks and measurements used in the study. Bizygomatic breadth was measured between the right and left zygion (zy–zy), morphological facial height from nasion to gnathion (n–gn), and morphological upper facial height from nasion to prosthion (n–pr).

Calculation of Facial Indices

The morphological facial index (MFI) was calculated as:

$$\text{MUFI} = \frac{\text{Morphological upper facial height}}{\text{Bizygomatic breadth}} \times 100$$

The morphological upper facial index (MUFI) was calculated as:

$$\text{MUFI} = \frac{\text{Morphological upper facial height}}{\text{Bizygomatic breadth}} \times 100$$

Statistical Analysis

Data were checked for completeness and consistency and analysed using JASP software version 0.98.1 developed in 2025. Continuous variables were assessed for normality using the Shapiro–Wilk test, supported by inspection of the distributions.Age

was non-normally distributed and was therefore summarised using the median and interquartile range and compared between groups using the Mann–Whitney U test. Sex was summarised as frequencies and percentages and compared using Pearson's chi-square test.

Bizygomatic breadth, morphological facial height, morphological upper facial height, morphological facial index and morphological upper facial index were approximately normally distributed and were summarised as mean ± standard deviation. Group differences were assessed using independent-samples Student's t-tests. Mean differences with 95% confidence intervals were reported, and Cohen's d was calculated to quantify effect size. All statistical tests were two-tailed, and statistical significance was set at $p < 0.05$.

RESULTS

Demographic Characteristics of the Study Participants

A total of 90 participants were included in the analysis, comprising 30 individuals with Down syndrome and 60 apparently healthy controls. The median age was 11.50 years (IQR 7.50) among participants with Down syndrome and 10.00 years (IQR 5.25) among controls. There was no statistically significant difference in age between the two groups ($U = 771.5$, $p = .272$). The Down syndrome group comprised 17 males (56.7%) and 13 females (43.3%), while the control group comprised 34 males (56.7%) and 26 females (43.3%). Sex distribution was identical proportionally between the groups and was not significantly different ($\chi^2 = 0.00$, $p = 1.000$).

Comparison of Craniofacial Anthropometric Measurements and Facial Indices

Bizygomatic breadth was slightly lower among participants with Down syndrome than among controls (118.4 ± 9.73 mm versus 121.5 ± 8.59 mm); however, the difference was not statistically significant (mean difference = 3.10 mm, 95% CI −0.89 to 7.09; $t(88) = 1.54$, $p = .126$). The corresponding effect size was small (Cohen's $d = 0.35$). Morphological facial height was significantly lower among participants with Down syndrome (98.67 ± 9.58 mm) than among controls (103.8 ± 9.02 mm), with a mean difference of 5.10 mm (95% CI 1.01–9.19; $t(88) = 2.48$, $p = .015$). The magnitude of this difference was moderate (Cohen's $d = 0.55$). Morphological upper facial height was also significantly lower in the Down syndrome group (54.87 ± 6.27 mm) compared with controls (59.40 ± 5.12 mm). The mean difference was 4.53 mm (95% CI 2.08–6.99; $t(88) = 3.67$, $p < .001$), corresponding to a

large effect size (Cohen's d = 0.82). A similar pattern was observed for the derived facial indices. The morphological facial index was significantly lower among participants with Down syndrome than controls (83.31 ± 3.92 versus 85.35 ± 3.12), with a mean difference of 2.04 (95% CI 0.53–3.56; t(88) = 2.69, p = .009; Cohen's d = 0.60). Morphological upper facial index demonstrated an even larger group difference, with values of 46.29 ± 2.91 in the Down syndrome group and 48.88 ± 2.25 among controls (mean difference = 2.59, 95% CI 1.48–3.69; t(88) = 4.65, p < .001; Cohen's d = 1.04).

Table 1. Demographic characteristics of participants

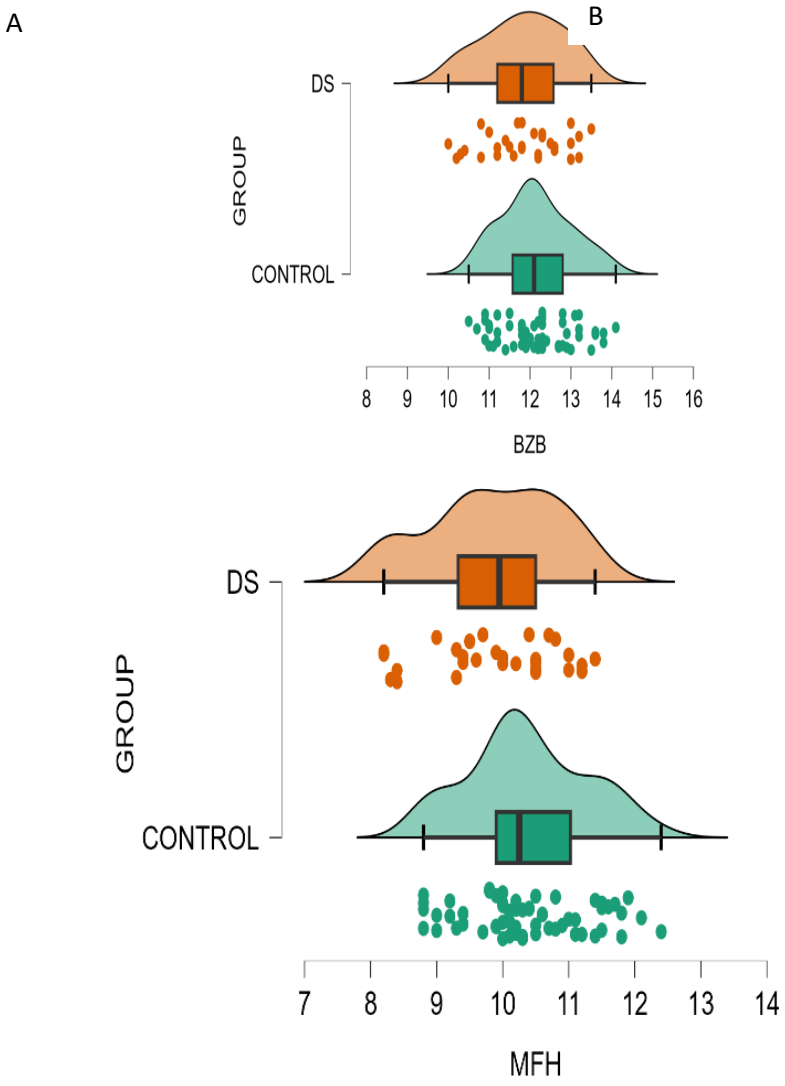
Characteristic	Control (n = 60)	Down syndrome (n = 30)	Test statistic	p-value
Age, years, median (IQR)	10.00 (5.25)	11.50 (7.50)	U = 771.5	.272
Sex, n (%)			$\chi^2 = 0.00$	p>.999
Female	26 (43.3)	13 (43.3)		
Male	34 (56.7)	17 (56.7)		

IQR = interquartile range; DS = Down syndrome. Age was compared using the Mann–Whitney U test, while sex distribution was compared using Pearson's chi-square test. The two groups did not differ significantly in age or sex distribution.

Table 2. Comparison of craniofacial anthropometric measurements and facial indices between participants with Down syndrome and controls

Variable	Control (n = 60), Mean ± SD	Down syndrome (n = 30), Mean ± SD	Mean difference (95% CI)	t(88)	p-value	Cohen's d
Bizygomatic breadth (mm)	121.5 ± 8.59	118.4 ± 9.73	3.10(−0.89 to 7.09)	1.54	.126	0.35
Morphological facial height (mm)	103.8 ± 9.02	98.67 ± 9.58	5.10 (1.01 to 9.19)	2.48	.015	0.55
Morphological upper facial height (mm)	59.40 ± 5.12	54.87 ± 6.27	4.53 (2.08 to 6.99)	3.67	<.001	0.82
Morphological facial index	85.35 ± 3.12	83.31 ± 3.92	2.04 (0.53 to 3.56)	2.69	.009	0.60
Morphological upper facial index	48.88 ± 2.25	46.29 ± 2.91	2.59 (1.48 to 3.69)	4.65	<.001	1.04

Values are presented as mean ± standard deviation. Independent-samples Student's *t*-tests were used for group comparison. CI = confidence interval; DS = Down syndrome. Facial indices are dimensionles


A
B
DS
CONTROL
GROUP
8 9 10 11 12 13 14 15 16
BZB
DS
CONTROL
GROUP
7 8 9 10 11 12 13 14
MFH

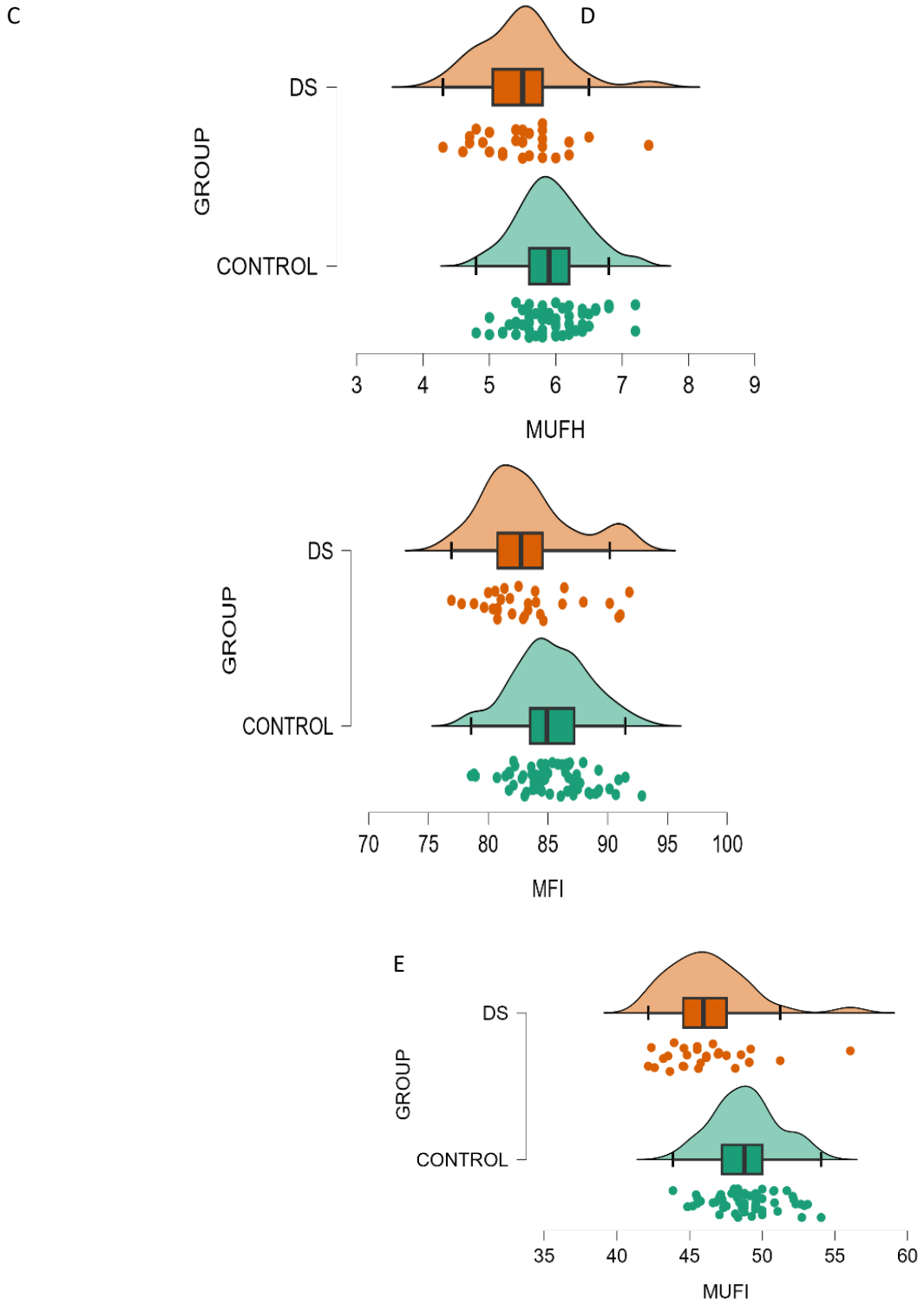


Figure 2. Distribution of craniofacial anthropometric measurements and facial indices among participants with Down syndrome and controls. Raincloud plots showing the distribution of (A) bizygomatic breadth (BZB), (B) morphological facial height (MFH), (C) morphological upper facial height (MUFH), (D) morphological facial index (MFI), and (E) morphological upper facial index (MUFI) in participants with Down syndrome and

controls. Each plot displays the distribution density, boxplot summary, and individual observations.

DISCUSSION

The present study compared selected craniofacial dimensions and facial indices between Nigerian individuals with Down syndrome and apparently healthy controls. The principal finding was that the craniofacial differences were more pronounced in the vertical facial dimensions and facial proportions than in transverse facial width. Although bizygomatic breadth was slightly lower among participants with Down syndrome, the difference was not statistically significant and the effect size was small. In contrast, morphological facial height and morphological upper facial height were significantly lower in the Down syndrome group, with moderate and large effect sizes, respectively. Similarly, both morphological facial index and morphological upper facial index were significantly lower among participants with Down syndrome, with the greatest standardised difference observed for the morphological upper facial index.

The absence of a statistically significant difference in bizygomatic breadth suggests that transverse facial width may be relatively preserved compared with the more pronounced alterations in vertical facial dimensions. This finding supports the view that craniofacial growth in Down syndrome is not uniformly reduced across all regions. Three-dimensional studies have demonstrated that different facial components are affected to varying degrees in individuals with Down syndrome, with alterations involving the maxillomandibular region and several other facial structures (Jayaratne et al., 2017). Interestingly, a recent Nigerian study also found no significant overall difference in morphological facial breadth between children with Down syndrome and controls, although the direction of the difference was not identical to that observed in the present study (Bashir et al., 2026). Earlier soft-tissue anthropometric investigations have likewise shown that some facial dimensions are substantially reduced whereas others demonstrate less pronounced differences, suggesting a region-specific rather than uniform effect of trisomy 21 on craniofacial morphology (Ferrario et al., 2004).

In contrast to bizygomatic breadth, morphological facial height was significantly reduced among individuals with Down syndrome in the present study. This finding is consistent with previous evidence demonstrating reduced vertical facial dimensions in

Down syndrome. In Kaduna State, Nigeria, Bashir et al. (2026) reported significantly lower morphological facial height among children with Down syndrome compared with controls. Similarly, Ferrario et al. (2004) demonstrated significantly shorter facial dimensions in individuals with Down syndrome compared with controls of comparable age and ethnicity. A systematic review and meta-analysis by Vicente et al. (2020) also reported significantly reduced anterior and posterior facial heights in individuals with Down syndrome, together with reductions in cranial base and maxillary dimensions. Although the latter studies used different anthropometric or cephalometric approaches, the consistency of these findings supports reduced vertical facial development as an important component of the Down syndrome craniofacial phenotype.

The reduction in morphological upper facial height was particularly notable in the present study, demonstrating a large effect size. This finding is anatomically consistent with the recognised involvement of the midfacial region in Down syndrome. Vicente et al. (2020) found reduced maxillary dimensions, reduced facial height and flattening of the middle third of the face in individuals with Down syndrome. Three-dimensional facial assessment has similarly demonstrated maxillomandibular hypoplasia and reduced dimensions of several facial structures in affected individuals (Jayaratne et al., 2017). There is also emerging developmental evidence for these morphological changes. Using experimental models of Down syndrome, Redhead et al. (2023) demonstrated that increased dosage of Dyrk1a and at least three other chromosome 21-related genes affects cranial neural crest-derived structures and reduces neural crest cell proliferation. Although these experimental findings cannot be directly equated with soft-tissue anthropometric measurements in humans, they provide biological plausibility for the altered craniofacial growth associated with trisomy 21.

The facial indices further demonstrate that the differences between the two groups involve facial proportionality as well as absolute dimensions. Morphological facial index was significantly lower in the Down syndrome group, while morphological upper facial index showed the largest effect size among the variables examined. Because these indices express vertical facial dimensions relative to bizygomatic breadth, their lower values indicate that the reduction in facial height is proportionally greater than any reduction in facial width. The resulting facial configuration is therefore relatively shorter vertically in relation to its transverse dimension. The importance of such proportional measures has previously been demonstrated by Asha et al. (2011), who found that morphological upper

facial index was among the craniofacial variables that could discriminate South Indian individuals with Down syndrome from unaffected controls. These findings support the value of examining facial indices alongside individual linear measurements when characterising the craniofacial phenotype of Down syndrome.

Population-specific interpretation is important in craniofacial anthropometry because facial dimensions and proportions vary according to population background as well as age and sex. In a study of young Nigerians, Olusanya et al. (2018) demonstrated that several facial proportions did not conform to classical neoclassical facial canons and advocated population-sensitive reference values for craniofacial assessment. More recently, three-dimensional stereophotogrammetric assessment of Nigerians demonstrated variations in facial dimensions according to sex and age (Adekunle et al., 2022). These observations underscore the importance of comparing individuals with Down syndrome with controls from the same population rather than relying exclusively on reference standards developed in other ethnic or geographical groups.

The demographic comparability of the groups strengthens the interpretation of the present findings. There was no statistically significant difference in age between participants with Down syndrome and controls, while the proportional distribution of males and females was identical in both groups. This is important because craniofacial dimensions vary with growth and may exhibit sexual dimorphism, including among Nigerian populations (Adekunle et al., 2022). Although residual effects of age and individual growth patterns cannot be completely excluded, the absence of major age and sex imbalance reduces the likelihood that these factors alone account for the observed group differences.

The findings have relevance to clinical anatomy, dysmorphology, orthodontics and craniofacial assessment. Direct anthropometry provides a relatively simple and non-invasive means of objectively quantifying facial morphology. Previous work has demonstrated the usefulness of craniofacial anthropometric profiles and indices for objectively characterising the Down syndrome phenotype (Asha et al., 2011; Ferrario et al., 2004). In the present study, the comparatively large differences in morphological upper facial height and morphological upper facial index suggest that measurements incorporating the upper facial region may be particularly useful descriptors of craniofacial morphology in this population. These measurements should, however, be regarded as quantitative descriptors rather than diagnostic tests in isolation.

Strengths and Limitations

A strength of the present study is the use of clearly defined anthropometric landmarks and a comparison group drawn from the same geographical population. The reporting of mean differences, confidence intervals and effect sizes also provides information on the magnitude of the observed differences beyond statistical significance alone. Nevertheless, some limitations should be considered. The relatively small number of participants with Down syndrome may limit the precision and generalisability of the findings. The cross-sectional design also prevents evaluation of individual craniofacial growth trajectories.

CONCLUSION

Overall, the findings indicate that the craniofacial phenotype of Down syndrome in this Nigerian population is characterised predominantly by reduced vertical facial dimensions and altered facial proportions rather than a marked reduction in bizygomatic breadth. The particularly pronounced differences in morphological upper facial height and morphological upper facial index are consistent with the recognised involvement of the midfacial region in Down syndrome. These findings provide population-specific quantitative data and support the usefulness of direct craniofacial anthropometry for objectively describing the facial phenotype of Nigerian individuals with Down syndrome.

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