Golestan University of Medical Sciences

Jorjani Biomedicine Journal

Online ISSN: 2645-3509

Digit ratio in autism spectrum disorder versus down syndrome: An anthropometric study in special needs schools of port Harcourt, Nigeria

Gwunireama Israel Ukie [1]*, Momodu Fawziyah Osinhe [1]

1. Department of Anatomy, Faculty of Basic Medical Sciences, University of Port Harcourt, Port Harcourt, Nigeria
* Correspondence: Gwunireama, Israel Ukie. Department of Anatomy, Faculty of Basic Medical Sciences, University of Port Harcourt, Port Harcourt, Nigeria. Tel: +2348033364860; Email: israel.gwunireama@uniport.edu.ng

Abstract

Background: The second-to-fourth digit ratio (2D:4D), obtained by dividing the length of the index finger by that of the ring finger, varies significantly by sex, race, and ethnicity. This ratio has previously been associated with both autism spectrum disorder and Down syndrome. The current study, therefore, examined the 2D:4D digit ratio in individuals with these two neurodevelopmental conditions recruited from special needs schools in Port Harcourt, Rivers State, Nigeria.

Methods: This cross-sectional descriptive study involved 200 participants: 100 with Down syndrome and 100 with autism. Participants met specific inclusion criteria, including the absence of hand deformities and a confirmed diagnosis. The lengths of the 2nd and 4th digits were measured with a digital vernier calliper. SPSS software, version 19, was used for data analysis. Statistical significance level was set at 0.05.

Results: Male autistic subjects showed a normal right-hand digit ratio (0.98 ± 0.12) but an elevated left-hand ratio (1.00 ± 0.01), whereas female autistic subjects displayed normal ratios bilaterally (1.00 ± 0.01 Left; 1.00 ± 0.00 Right). Males with Down syndrome exhibited higher-than-normal ratios on both hands (Left: 1.02 ± 0.03; Right: 1.01 ± 0.01), and females showed ratios of 1.01 ± 0.01 (Left) and 1.00 ± 0.00 (Right). Mean comparisons revealed significant differences (P-Value < 0.05) between autistic and Down syndrome subjects of both sexes, with individuals with Down syndrome consistently demonstrating higher digit ratios than autistic individuals.

Conclusion: Subjects with Down syndrome had significantly higher digit ratios than autistic subjects. This finding confirms that digit ratios are sexually dimorphic and suggests hypermasculinity in autistic males because of their lower digit ratios.

Article Type: Brief Report

Article History

Received: 5 February 2024
Received in revised form: 12 March 2025
Accepted: 10 April 2025
Available online: 28 December 2025
DOI: 10.29252/jorjanibiomedj.13.4.23

Keywords

Digit Ratio
Autistic Disorder
Down Syndrome

Highlights

What is current knowledge?

Autism and Down syndrome influence digit lengths and, therefore, affect the 2D:4D ratio.

What is new here?

Subjects with Down syndrome have higher digit ratios than autistic subjects.

Introduction

The digit ratio, often called 2D:4D, represents the proportional relationship between the lengths of the index finger (2D) and the ring finger (4D). The index finger is the second finger from the thumb, and the ring finger is the fourth finger, located between the middle and little fingers (1,2). This ratio has been widely studied as a non-invasive biomarker, offering insights into prenatal hormonal influences and serving as a potential predictor of susceptibility to various diseases and developmental conditions, such as immune dysfunction, myocardial infarction, and breast cancer. Low 2D:4D digit ratios, in which the second finger is shorter than the fourth, indicate higher fetal testosterone compared with estrogen. Conversely, high digit ratios, in which the second finger is longer than the fourth, suggest higher fetal estrogen relative to testosterone (3,4). The right side of the body shows the most pronounced expression of these sexually dimorphic traits (5).

Digit ratios show interesting differences between sexes, known as sexual dimorphism. Males generally exhibit lower digit ratios, with an average of 0.98, whereas females average around 1.00. This variation is connected to the developmental relationship between the urogenital system and fingers, both of which are influenced by Hox genes. Evidence suggests a positive relationship between male finger length ratios and various traits influenced by testosterone during adolescence and adulthood. Understanding these relationships could provide valuable insights into developmental biology and human behaviour (6-8).

Autism spectrum disorder (ASD), commonly known as autism, describes a group of developmental brain irregularities. Approximately 1 in 160 children worldwide are affected by ASD, which equates to a prevalence rate of 62 per 10,000 children. The exact cause of autism remains unknown. It is highly heritable and is believed to be primarily genetic; however, many genes are involved, and environmental factors may also play a significant role (9-11). Higher prenatal testosterone levels are linked to the development of ASD traits. Because low digit ratios correspond to increased prenatal testosterone, these ratios are proposed as indicators of a greater likelihood of exhibiting autistic characteristics (12). Researchers have described autism as an extreme form of certain sexually dimorphic characteristics, commonly explained by the “extreme male brain” hypothesis. This theory suggests that autism results from an exaggerated male brain pattern, a condition sometimes called hypermasculinisation, which is observed in autistic children. Because of this, the digit ratio may serve as a useful biomarker indicating the likelihood of developing autistic traits. Research supports that autistic boys and girls tend to show signs of hypermasculinisation, such as increased facial masculinity, which correlates with autism-related social communication difficulties. Thus, the digit ratio could reflect prenatal hormonal influences linked to these autistic characteristics (13-15).

Down syndrome, also known as trisomy 21, is a genetic disorder defined by the presence of an additional full or partial copy of chromosome 21. This additional chromosome can influence various aspects of a person’s development, including physical traits and intellectual capabilities (16). Research has revealed interesting patterns in digit ratios (2D:4D) among individuals with Down syndrome. Specifically, males tend to exhibit higher ratios, while females show lower ratios, contrasting with the typical trends observed in the general population (17,18).

This study aimed to investigate and compare the digit ratio (2D:4D) among individuals with autism spectrum disorder (ASD) and Down

syndrome (DS) in special needs schools in Port Harcourt, Rivers State, to identify potential differences in digit ratio patterns and explore their implications for understanding developmental and genetic influences in these populations.

Methods

This study used a cross-sectional descriptive design involving a total of 200 participants selected from three special needs schools in Port Harcourt, Rivers State, Nigeria. Participants were enrolled using a convenience sampling method. Among the participants, 100 were diagnosed with Down syndrome (60 males, 40 females; age range, 3 - 23 years), and another 100 had autism (55 males, 45 females; age range, 2 - 27 years). The careful selection and recruitment process aimed to create a representative sample that would enhance understanding of both conditions and inform future research and interventions.

The inclusion criteria for the study were as follows: participants had to be diagnosed with either autism or Down syndrome, be at least 2 years old, and have no hand deformities, such as polydactyly or oligodactyly, or evidence of trauma on the second or fourth digits. The exclusion criteria consisted of the presence of congenital or acquired hand or finger deformities, a history of hand surgery or trauma, and inability to cooperate with finger measurement. Informed consent was secured from the parents or guardians of all participants before they joined the study. Each participant was clearly informed about the research aims, characteristics, and procedures, with information delivered in simple and plain language. Participants were informed that they could leave the study whenever they chose without any restrictions. This ensured respect for their autonomy and freedom throughout the research process. The process was designed to align with ethical guidelines for research involving human subjects, demonstrating a commitment to respecting participants' autonomy and rights.

All anthropometric measurements, including finger length and digit ratio (2D:4D), were performed by a single trained evaluator who was blinded to the participants' diagnostic groups (Autism or Down syndrome) to minimize measurement bias. The evaluator had a background in physical anthropology and received standardized training in finger landmark identification and digital calliper use before data collection. The lengths of the second and fourth digits were measured from the basal crease to the fingertip using a digital vernier calliper, allowing precise measurements to the nearest 0.01 mm. Each digit was measured twice to ensure accuracy, and the average of these measurements was used as the final recorded value. Subsequently, digit ratios were calculated for each subject across both hands. Data analysis was performed using SPSS software, version 19.0. For each hand, descriptive statistics (Mean, standard deviation, and range) of the 2D:4D digit ratios were calculated separately for the autism group and the Down syndrome group. The Shapiro-Wilk test was applied to check for normal distribution. An independent t-test was used to compare the groups' mean digit ratios if the data were normally distributed; otherwise, the Mann-Whitney U test was used. The statistical significance level was set at 0.05.

Results

For the autistic subjects, the descriptive results showed that males had a digit ratio of less than 1.00 (0.98 ± 0.12) on the right hand, which is a normal digit ratio for males. However, the digit ratio for males on the left hand was 1.00 ± 0.01, which was high for males. The female autistic subjects had digit ratios of 1.00 ± 0.01 and 1.00 ± 0.00 on the left and right hands, respectively. These values were normal digit ratios for females (Table 1).

Male subjects with Down syndrome had digit ratios of 1.02 ± 0.03 on the left hand and 1.01 ± 0.01 on the right hand. Female subjects with Down syndrome had digit ratios of 1.01 ± 0.01 and 1.00 ± 0.00 on the left and right hands, respectively (Table 2).

The results of the mean comparison of measured variables between male autistic and Down syndrome subjects showed significant differences (P-Value < 0.05) in digit ratios between male autistic and Down syndrome patients. Additionally, negative t-values were observed for the left and right digit ratios, approximately -2.61 and -4.17, respectively (Table 3).

The negative t-values indicate that male subjects with Down syndrome had significantly higher digit ratios than male autistic subjects, because the Down syndrome group showed means of 1.02 on the left hand and 1.01 on the right hand, while autistic males showed 0.98 and 1.00, respectively.

An independent t-test revealed statistically significant differences (P-Value < 0.05) across all measured parameters when comparing female subjects with autism to female subjects with Down syndrome (Table 4). Additionally, the results showed that subjects with Down syndrome had higher digit ratios than autistic subjects.

Table 1. Descriptive statistics of the measured variables in male and female autistic subjects

Measured variables	Male [N = 55]			Female [N = 45]			Total [N = 100]		
	Min	Max	Mean ± SD	Min	Max	Mean ± SD	Min	Max	Mean ± SD
Age (Years)	4.00	22.00	11.22 ± 4.72	2.00	27.00	11.91 ± 6.96	2.00	27.00	11.53 ± 5.81
Left 2D	3.40	8.95	5.54 ± 1.39	2.51	9.88	5.79 ± 1.82	2.51	9.88	5.65 ± 1.60
Left 4D	3.41	8.60	6.15 ± 1.27	2.51	9.88	5.80 ± 1.82	2.51	9.89	5.99 ± 3.38
Left 2D:4D	0.10	1.02	0.98 ± 0.12	0.97	1.00	1.00 ± 0.01	0.10	1.02	0.99 ± 0.09
Right 2D	3.41	8.97	5.60 ± 1.38	2.51	9.88	5.79 ± 1.82	2.51	9.88	5.69 ± 1.59
Right 4D	3.41	8.97	5.57 ± 1.39	2.51	9.87	5.79 ± 1.82	2.51	9.87	5.67 ± 1.59
Right 2D:4D	1.00	1.03	1.00 ± 0.01	1.00	1.00	1.00 ± 0.00	1.00	1.03	1.00 ± 0.01

Min = Minimum, Max = Maximum, SD = Standard Deviation, N = Number of subjects, D = Digit

Table 2. Descriptive statistics of the measured variables in male and female down syndrome subjects

Measured variables	Male [N = 60]			Female [N = 40]			Total [N = 100]		
	Min	Max	Mean ± SD	Min	Max	Mean ± SD	Min	Max	Mean ± SD
Age (Years)	3.00	22.00	9.18 ± 4.49	4.00	23.00	10.63 ± 4.98	3.00	23.00	9.76 ± 4.72
Left 2D	2.75	6.90	4.23 ± 0.98	3.17	6.87	4.20 ± 0.99	2.75	6.90	4.22 ± 0.98
Left 4D	2.69	6.81	4.15 ± 0.98	3.15	6.84	4.17 ± 0.99	2.69	6.84	4.16 ± 0.98
Left 2D:4D	0.99	1.22	1.02 ± 0.03	1.00	1.04	1.01 ± 0.01	0.99	1.22	1.02 ± 0.02
Right 2D	2.77	6.90	4.25 ± 0.97	3.20	6.87	4.23 ± 0.99	2.77	6.90	4.25 ± 0.97
Right 4D	2.76	6.81	4.21 ± 0.96	3.18	6.87	4.21 ± 0.99	2.76	6.87	4.21 ± 0.97
Right 2D:4D	1.00	1.04	1.01 ± 0.01	1.00	1.02	1.00 ± 0.00	1.00	1.04	1.01 ± 0.01

Min = Minimum, Max = Maximum, SD = Standard Deviation, N = Number of subjects, D = Digit

Table 3. Mean comparison of the measured variables in male autistic and down syndrome subjects using independent t-test

Measured variables	MD	SE	95% C.I of the difference		df	t-value	P-Value
			Lower	Upper			
Age (Years)	2.03	0.86	0.33	3.74	113.00	2.37	**0.02***
Left 2D	1.31	0.23	0.86	1.76	96.05	5.78	**< 0.001***
Left 4D	2.00	0.59	0.82	3.18	59.21	3.39	**< 0.001***
Left 2D:4D	-0.04	0.02	-0.07	-0.01	113.00	-2.61	**0.01***
Right 2D	1.34	0.22	0.90	1.79	95.69	6.00	**< 0.001***
Right 4D	1.37	0.22	0.92	1.81	94.98	6.09	**< 0.001***
Right 2D:4D	-0.01	0.00	-0.01	0.00	113.00	-4.17	**< 0.001***

* = Significant at P-Value < 0.05, MD = Mean Difference, SE = Standard Error, CI = Confidence Interval, df = Degree of Freedom

Table 4. Mean comparison of the measured variables in female autistic and down syndrome subjects using independent t-test

Measured variables	MD	SE	95% C.I of the difference		df	t-value	P-Value
			Lower	Upper			
Age (Years)	1.29	1.30	-1.31	3.88	79.53	0.99	**0.33**
Left 2D	1.58	0.31	0.96	2.21	69.50	5.05	**< 0.001***
Left 4D	1.63	0.31	1.01	2.26	69.66	5.21	**< 0.001***
Left 2D:4D	-0.01	0.00	-0.02	-0.01	62.11	-7.84	**< 0.001***
Right 2D	1.56	0.31	0.93	2.18	69.54	4.98	**< 0.001***
Right 4D	1.58	0.31	0.95	2.20	69.38	5.03	**< 0.001***
Right 2D:4D	0.00	0.00	-0.01	0.00	43.83	-7.07	**< 0.001***

* = Significant at P-Value < 0.05, MD = Mean Difference, SE = Standard Error, CI = Confidence Interval, D = Digit

Discussion

The present study compared the digit ratios (2D:4D) between autistic and Down syndrome subjects of both sexes. Descriptive findings revealed that male autistic subjects exhibited a normal right digit ratio but a slightly lower left digit ratio compared with typical male norms, while female autistic subjects showed normal female digit ratios bilaterally. In contrast, male subjects with Down syndrome demonstrated higher-than-normal digit ratios on both hands, and female subjects with Down syndrome showed slightly elevated digit ratios compared with typical female values. Inferential statistical analysis indicated that, for both males and females, subjects with Down syndrome had significantly higher digit ratios than autistic subjects across both the left and right hands. These differences were statistically significant for all comparisons.

Previous studies have established that males usually have a higher fourth digit length and a lower second digit length, resulting in a digit ratio of less than 1.00 and, therefore, a low digit ratio. However, females have a higher second digit length and a lower fourth digit length, resulting in a digit ratio of ≥ 1.00 and, therefore, a high digit ratio (19-21). In the present study, female autistic subjects showed a higher digit ratio than their male counterparts, which agrees with the established findings of other researchers (19-21). However, the results for subjects with Down syndrome contrasted with the established pattern of sexual dimorphism in digit ratio (19-21). This was because male subjects with Down syndrome had a digit ratio > 1.00. The present study therefore highlights a key distinction between the two neurodevelopmental conditions regarding sexual dimorphism of digit ratio. While autistic subjects preserved the typical sex difference (Males < 1.00, females ≥ 1.00), subjects with Down syndrome exhibited a disrupted or reversed pattern, with males showing feminized ratios (> 1.00). This suggests that the prenatal hormonal environment may be differentially affected in these two conditions. Autism appears to maintain normative androgen-mediated sexual differentiation, whereas Down syndrome may involve an atypical hormonal profile characterized by relatively lower fetal testosterone exposure in males. The findings emphasize that digit ratio could serve as a simple biomarker for identifying hormonal imbalances during critical stages of development in neurodevelopmental disorders.

In the present study, male subjects with Down syndrome had digit ratios of 1.01 ± 0.01 and 1.02 ± 0.03 on the right and left hands, respectively. In females, the digit ratio was 1.00 ± 0.00 on the right side and 1.01 ± 0.01 on the left side. This is noteworthy because a high digit ratio reflects higher fetal estrogen levels in relation to fetal testosterone levels (2,4). Therefore, the results of this study suggest that male subjects with Down syndrome may be exposed to high fetal estrogen and low fetal testosterone levels, resulting in a high digit ratio ≥ 1.00. Research conducted by Viveka et al. (18), which examined digit ratio and perinatal androgen exposure in Down syndrome, revealed that females had notably lower digit ratios than males, contrasting with findings in normal populations. This finding supports the results from both Suresh et al. (17) and the current investigation.

Taken together, these findings suggest that Down syndrome is associated with a masculinized digit ratio pattern in females (i.e., lower ratios than typical female norms) and a feminized pattern in males (i.e., higher ratios ≥ 1.00). This reversal of the typical sex difference in digit ratio indicates that the prenatal hormonal milieu in Down syndrome may be characterized by relatively lower androgen exposure in males and relatively higher androgen exposure in females compared with their typically developing counterparts. The consistency between the present study and previous reports by Suresh et al. (17) and Viveka et al. (18) strengthens the evidence that altered prenatal steroid hormone activity may play a role in the phenotypic expression of Down syndrome.

The present study found that individuals with Down syndrome had digit ratios of 1.01 ± 0.01 on the right hand and 1.02 ± 0.02 on the left hand. These values were higher than those observed in autistic individuals, who showed digit ratios of 1.00 ± 0.01 (Right) and 0.99 ± 0.09 (Left). An independent t-test indicated significant differences (P-Value < 0.05) between the right and left digit ratios of male participants with autism and those with Down syndrome. Moreover, significant differences in both right and left digit ratios were also found between the two diagnostic groups overall.

Conclusion

The findings of this study demonstrate that the digit ratio (2D:4D) differs significantly between individuals with autism spectrum disorder and those with Down syndrome. Autistic subjects, particularly males, exhibited digit ratios closer to typical sex-specific norms, whereas subjects with Down syndrome showed consistently higher digit ratios in both sexes. These results suggest that prenatal hormone exposure, as

reflected by digit ratio, may differ between the two conditions. The higher digit ratios observed in Down syndrome may indicate relatively lower prenatal androgen exposure compared with autistic individuals. This study supports the potential use of digit ratio as a simple, non-invasive anthropometric marker for differentiating neurodevelopmental conditions. However, further investigations using larger cohorts and longitudinal designs are recommended to confirm these observations and explore the hormonal mechanisms that may explain them.

Acknowledgement

The authors would like to express their heartfelt gratitude to all the volunteers who participated in this research and to the parents and guardians who graciously provided their consent. Their involvement is crucial to advancing understanding in this field. Furthermore, the authors appreciate the dedicated staff at Otana School for Hearing and Edu-Health Services, Utmost Care and Therapy Services, and OPM Special Needs School for their essential support, which greatly enriched this study. Thank you for contributing to this important work.

Funding sources

This study did not receive any financial support or grants from funding organizations or agencies.

Ethical statement

This study successfully received ethical clearance and was approved by the University of Port Harcourt ethical clearance board, ensuring that all research practices aligned with ethical standards.

Conflicts of interest

There is no conflict of interest.

Author contributions

All authors participated in different aspects of the study, including designing the research, conducting the work, performing statistical analyses, and writing the manuscript.

Data availability statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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Cite this article as:

Gwunireama I.U, Momodu FO. Digit ratio in autism spectrum disorder versus down syndrome: An anthropometric study in special needs schools of port Harcourt, Nigeria. *Jorjani Biomedicine Journal.* 2025;13(4):23-6. http://dx.doi.org/10.29252/jorjanibiomedj.13.4.23