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ORIGINAL ARTICLE

Anthropometric Estimation of Metacarpophalangeal Profile in Individuals with Brachydactyly: A Radiometric Study

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ABSTRACT

Background: The bone length of paediatric patients varies with respect to normal individuals. Patients were mainly affected with congenital autopod anomalies such as brachydactyly, syndactyly, and polydactyly. Brachydactyly with short bones are further divided into different types: Brachydactyly A, brachydactyly B, brachydactyly C, brachydactyly D, and brachydactyly E.

Aim: This study aims to classify the types of brachydactyly by measuring metacarpophalangeal profile patients: A radiometric study

Objectives: Classify the types of brachydactyly patients and compare them with the controls based on X-rays of hands.

Material: A cross-sectional study was conducted on the children at a tertiary care center from July 2020 to March 2024. Metacarpal and phalangeal lengths of little, ring, middle, index, and thumb fingers on X-ray films of patients and controls were measured by digimatic calipers. The recorded data were analysed by SPSS version 26 and Graph Pad Prisma 8.2.

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Result: We found highly significant p values in ratios of phalangeal lengths 2P:3P, 2P:4P, 2P:5P, 2L:3L, 2L:4L, 2L: 5L (P; Proximal phalanx length , L: middle phalanx length). This predicted abnormal or short/missing proximal, middle, and distal phalanges of individuals with brachydactyly type A. We found highly significant p values in ratios of (MC: Metacarpal length) 2MC: 3MC, 2MC:4MC, 2MC:5MC, and 2P:4P in brachydactyly type E.

Conclusion: This predicted short 3rd, 4th, 5th metacarpals and 4th proximal phalanges in comparison to controls. Thus, X-ray analysis is the gold standard tool for evaluating the different types of brachydactyly. The research emphasizes the importance of hand anthropometry in understanding anatomical changes and the diversity of brachydactyly.

KEYWORDS

• Acral shortening • Brachymetacarpy • Digimatic Caliper • Fingers • Hand Anomalies • Human diversity • Metacarpals • Phalanges

Key Message: This study helps to classify the rare genetic disease in pediatric patients and better understanding for the genetic counselling. Also helpful in the surgical treatment of hands in complex cases.

INTRODUCTION

Background: Congenital autopod anomalies can occur as isolated malformations and in combination with other malformations of the stylopod, zygopod, or as part of a genetic syndrome. The mainly found congenital autopod malformations are brachydactyly, syndactyly, polydactyly.[1] In 2001, 'Brachydactyly' was added to the "International Nosology and Classification of Genetic Skeletal Disorders.[2] The brachydactyly groups are isolated and syndromic also classified in the "Nosology of genetic skeletal disorders" 11th revision and now contains 771 entries associated with 552 genes.[3] Brachydactylyis an autosomal dominant inherited trait. The upper limb is more frequently affected than the lower limb. Brachydactyly can exist in an isolated form or as a part of a syndrome. It is classified as brachydactyly A (BDA), brachydactyly B (BDB), brachydactyly C (BDC), brachydactyly D (BDD) and brachydactyly E (BDE).[4,5] In BDA patient has missing middle phalanges, or short middle, proximal, and distal phalanges. BDB is characterized by the absence or incomplete developmental growth of the distal and middle phalanx of all fingers of the hands and feet. Nail dysplasia is present in all fingers or sometimes shown in the 2nd and 5th digit. BDC is more complex type in all types of brachydactyly. It shows brachymesophalangy of the 2 and 3 digits and a shortened first metacarpal of the thumb. In BDD only the distal phalanx of the thumb is found shortened. BDE does not affect the phalanges but is majorly seen in metacarpals and metatarsals.[6] The shortened metacarpals and metatarsals are reflected in abnormal sizes of fingers and toes. Most individuals show 4th and 5th metacarpals and metatarsal shortening. The most common gene variation found in the different types of brachydactyly include in *IHH, HOXD13, GDF5,* and *PTHLH.*[7,8]

Syndactyly is mainly found in isolated cases and complex syndromes. The syndromes with severe brachy-syndactyly are Apert syndrome, Saethre Chotzen syndrome, Crouzon syndrome, and Pfeiffer syndrome. Syndactyly can exist as a cutaneous form or fusion of 3rd 4th and 5th digits. Polydactyly is commonly found in isolated forms as pre-axial and post-axial types.[9,10]

In a different populations, the morphometric parameters of the hand show a considerable degree of dimorphism. Few studies used to compare the normal population limb measurements of hand length, hand breadth, finger length, metacarpals length, and stature.[11,12] 2D:4D ratio was measured from photocopies and X-rays of hands in the rural Jamaican children. Studies also suggest the 2D:4D ratio may be associated with the expression of other sexually dimorphic behavioral traits.[13] Literature is scarce on congenital brachydactyly metacarpals and phalangeal length for isolated and syndromic

patients. This study was primarily aimed to classify the congenital autopod anomalies of BDA, and BDE based on the length of the metacarpals, proximal, middle, and distal phalanges on X-ray.

MATERIAL AND METHODS

The study was designed as a cross-sectional descriptive cohort study conducted on 62 individuals. Out of 62 individuals, 31 brachydactyly patients and 31 controls of the same age and gender were included. All the patients and controls are taken from the genetic clinic and general paediatric outpatient clinic of a tertiary care center. The patients and controls were enrolled from July 2020 to March 2024. All the X-rays of the hands of patients were conducted with standard equipment from Samsung Electronics Co. Ltd, Korea, GC85A, G-XR-190239. The standard distance of the detector from the object was 100cm. Hand metacarpals and phalangeal length were measured on X-Ray films using a horizontal viewing board in the Growth Lab, by digimatic sliding caliper (Least count: 0.01mm, make: Mitutoyo, Japan). The Institute Ethics Committee (IEC) clearance (ID: NK/7963/PhD/638) was taken before the enrolment of individuals in the study. This study enrolled isolated and syndromic patients with congenital autopod anomalies with appropriate written informed consent.

Inclusion Criteria: Individuals with congenital autopod anomalies. The age group of patients included is 7-34 years old both males and females.Isolated and also familial cases of brachydactyly were included. Controls were unaffected age and sex-matched individuals.

Exclusion Criteria: Patients with Down syndrome, Apert syndrome, Turner Syndrome, and definitive diagnosis of skeletal dysplasia were excluded. Those with accidental, old fracture in the hand were excluded. Patients with known endocrine, and suspected lysosomal storage disorders were not included.

Measurements on X-ray films

Both hand's x-ray films, metacarpals, and phalangeal length were measured in millimetres (mm) by a Digimatic sliding caliper on a horizontal viewing board at the growth lab. Metacarpals were taken as 1MC, 2MC, 3MC, 4MC, 5MC, Proximal phalanges taken as 1P, 2P, 3P, 4P, 5P; Middle phalanges taken as 2L, 3L, 4L, 5L; Distal phalanges taken as 1D, 2D, 3D, 4D, 5D showed in *figure 1*.

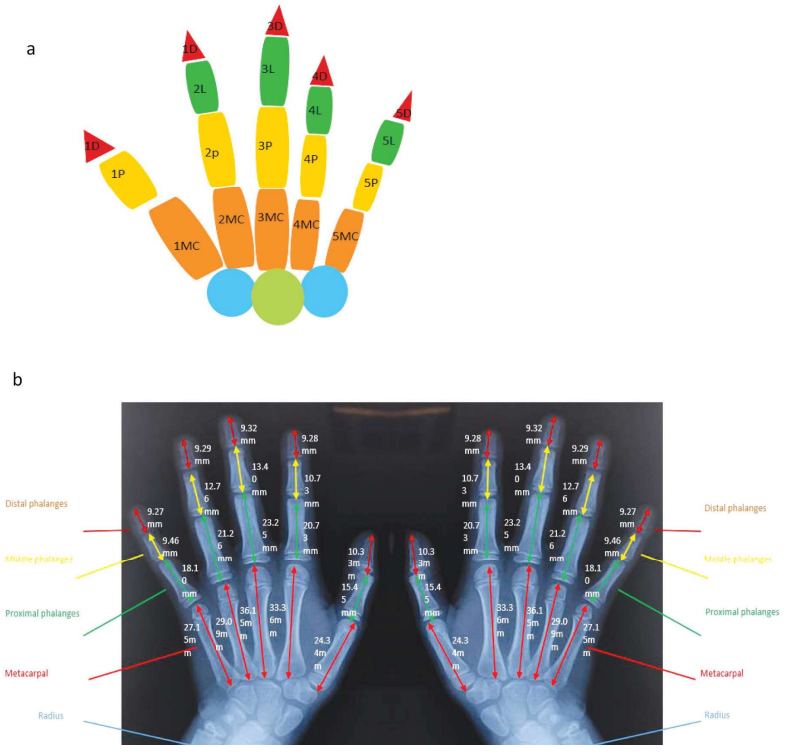

Figure 1: X-ray film measurements taken by Digimatic caliper:
A metacarpals and phalanges numbering, **B** Measurements on X-ray by Callipers

The standard parameters for measuring the length of metacarpals and phalanges are listed in *table 1*. Being least affected, index finger 2MC and index finger phalanges (2P, 2L, 2D) were taken as standard for estimating ratio with all other phalanges and metacarpals. The standard estimation ratio also used in previous literature. The ratio of metacarpals to metacarpals is 2MC:1MC, 2MC:3MC, 2MC:4MC, 2MC:5MC. The ratio for proximal phalanges length as 2P:1P, 2P:3P, 2P:4P, 2P:5P. For middle phalanges length ratio were calculated as 2L:1L, 2L:3L, 2L:4L, and distal phalanges as 2D:1D, 2D:3D, 2D:4D, 2D:5D.

Table 1: Standard parameters for measuring the length of metacarpals and phalanges

Parameters	Measurement observation
Metacarpal length (MC)	Measured the distance between the tip of the proximal phalanx to the elongated base of metacarpals. 1MC: Thumb; 2MC: Index finger; 3MC: Middle finger; 4MC: Ring finger; 5MC: little finger
Proximal phalanx length (P)	Measured the distance between the elongated tip of the proximal phalanx to the base of the distal phalanx. 1P: Thumb; 2P: Index finger; 3P: Middle finger; 4P: Ring finger; 5P: Little finger
Middle phalanx length (L)	Measured the distance between the base of the middle phalanx to the elongated tip of the middle phalanx. 2L: Index finger; 3L: Middle finger; 4L: Ring finger; 5L: Little finger
Distal Phalanx length (D)	Measured distance from the elongated tip of the distal phalanx to the elongated base of the distal phalanx. 1D: Thumb; 2D: Index finger; 3D: Middle finger; 4D: Ring finger; 5D: Little finger

Statistical analysis

The data was collected in MS Excel and analysed using SPSS software version 26 and Graph Pad PRISMA 8.2. The normality of the data was testing using Q-Q plot and the data with found to be normally distributed. The results for continuous variables were recorded as Mean ±SD. The difference between the mean values of the two groups was performed using an unpaired t-test. For interpretation of results of significance, non-significance was adopted at $P>0.05$; significance was adopted at $P< 0.05$, and high significance was taken at $P < 0.001$.

RESULTS

This study enrolled 62 individuals including 31 individuals with brachydactyly and 31 healthy controls. X-rays showing missing middle phalanges in BDA is depicted in *figure 2*. The x-ray films showing short 3rd 4th metacarpals in BDE as depicted in figure 3. Distribution of 62 patients and controls male and females showed in *Figures 4-5*. The study subjects and controls enrolled in the study were from different geographical urban and rural regions of India namely, Chandigarh, Punjab, Himachal Pradesh, Haryana, Jammu Kashmir, Uttar Pradesh, and Bihar. Metacarpals and phalangeal length were measured after obtaining the X-ray of both hands, followed by calculating the ratio of 2MC metacarpal to the other metacarpals 2MC:1MC, 2MC:3MC, 2MC:4MC, 2MC:5MC, 2P proximal phalanx to the other proximal phalanges, 2P: 1P, 2P:3P, 2P:4P, 2P:5P, 2L middle phalanx to the other middle phalanges 2L:3L, 2L:4L, 2L:5L, 2D distal phalanx to the other distal phalanges 2D:1D, 2D:3D, 2D:4D, 2D:5D.

We enrolled 14 individuals with BDA based on phenotype and also were evaluated the genotype by whole exome sequencing and Sanger sequencing. In 14 BDA patients, we have 4 affected individuals of the same family who showed autosomal dominant inheritance, the rest belonged to unrelated families. The study subjects had anomalies of missing middle phalanges, short middle phalanges, short proximal phalanges, and short stature. The patients BMI, height for age and weight for age were predicted as short stature. Out of 14 patients, 10 were found with short stature out of which 5 were females and 5 were males. Detailed discerption about the phenotype is showing in supplementary table 1S.

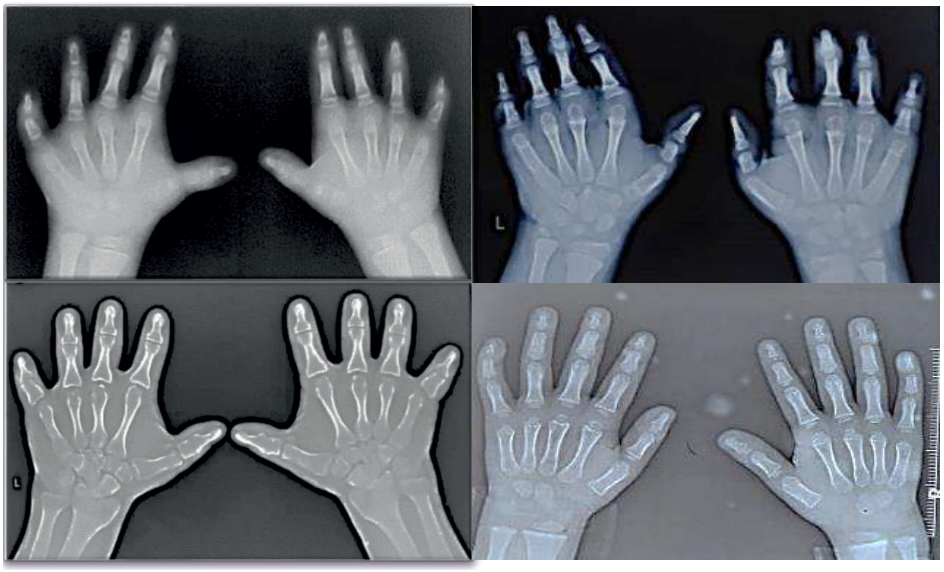

Figure 2: X-ray film of affected individuals of BDA

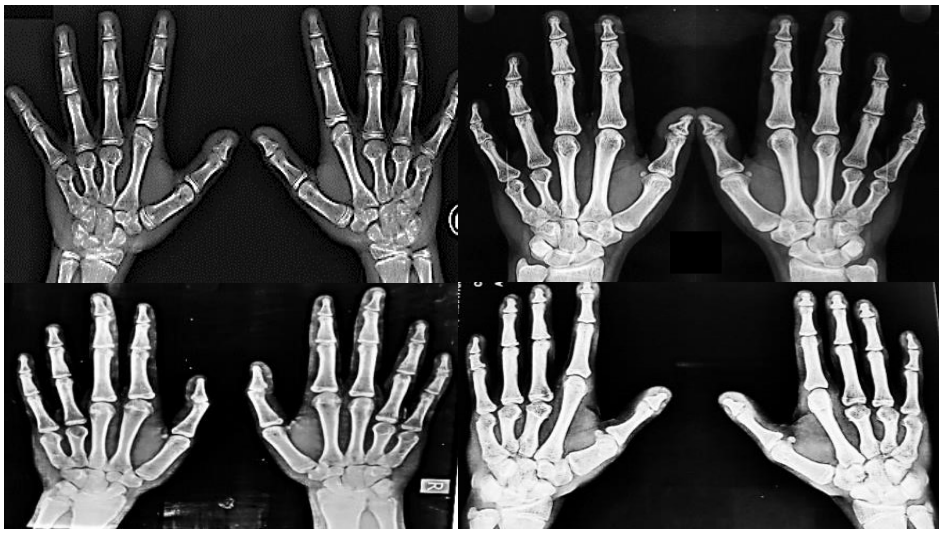

Figure 3: X-ray film of affected individuals of BDE

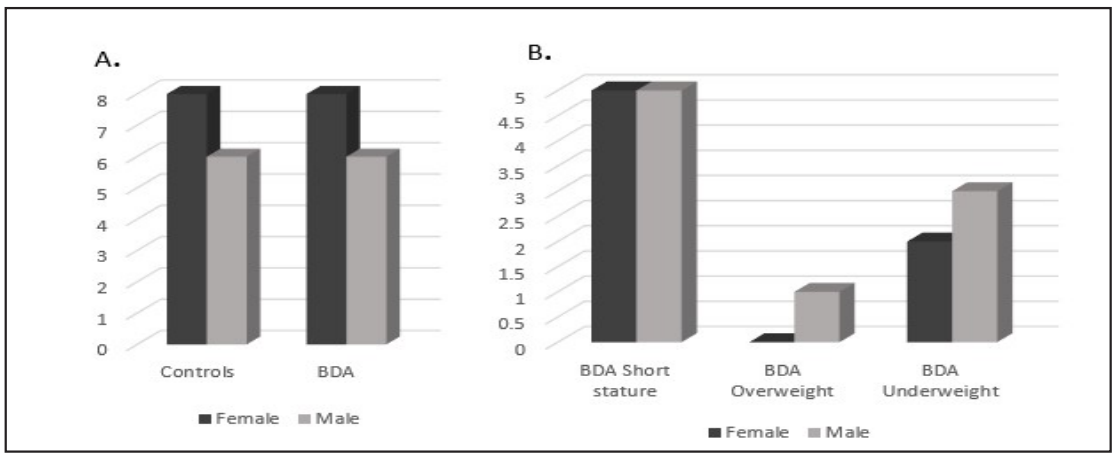

Figure 4: A. Distribution of controls and BDA patients, **B.** distribution of BDA patients on the bases of male and female short stature, overweight, and underweight

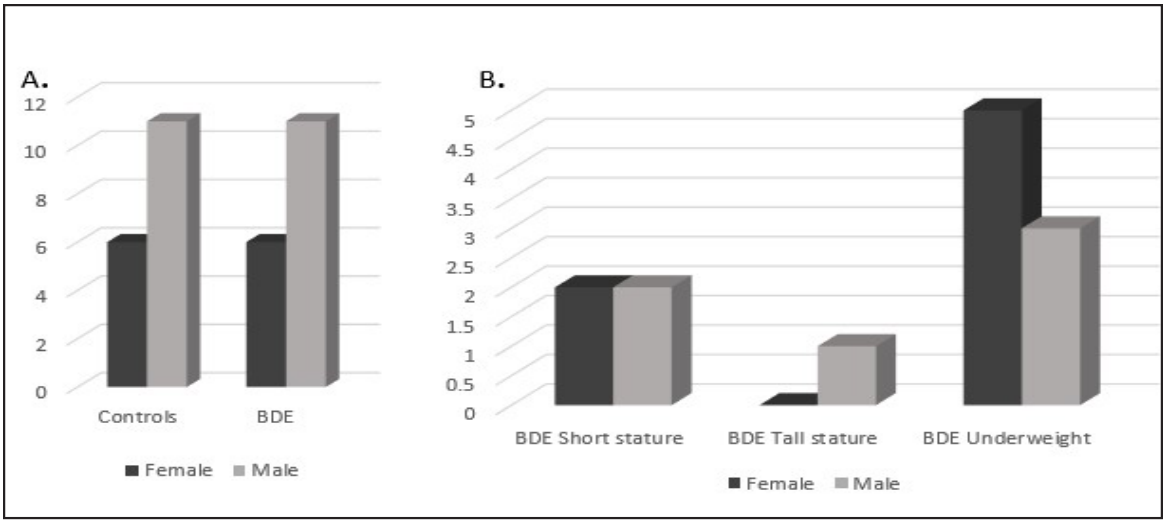

Figure 5: A. Distribution of controls and BDE patients,
B. distribution of BDE patients on the bases of male and female short stature, overweight, and underweight

Table 1: Phenotypic appearance of BDA patients, interpret non-syndromic and syndromic (NS/S) patients, BMI, height for age, weight for Age

Patients ID's	NS/S	Age/ Gender	Upper linb	Lower Limb	BMI	Height for age (HAZ)	Height	Weight for Age (WAZ)	Weight
BDA-1	NS	7YM	Short hands	Short feet	17.36	–0.81	Normal stature	0.48	Healthy Weight
BDA-2	NS	10YF	Short hands	—	13.9	–2.72	Short stature	–1.54	Healthy Weight
BDA-3	S	18YF	Short hands	Short feet	14.6	–.87	Normal stature	–2.54	Under Weight
BDA-4	NS	8YF	Short hands	Short feet	16.83	–3.65	Short stature	–1.56	Healthy Weight
BDA-5	S	8YM	Short hands	Short feet	15.1	–3.45	Short stature	–1.99	Healthy Weight
BDA-6	NS	7YF	Missing middle phalange of hands	—	13.16	–3.65	Short stature	–2.62	Under Weight
BDA-7	NS	10.3YF	Missing middle phalange of hands	—	14.8	–3.08	Short stature	–1.99	Healthy Weight
BDA-8	NS	37YM	Missing middle phalange of hands	—	25.2	2.01	Normal stature	2.22	Over Weight
BDA-9	NS	34YF	Missing middle phalange of hands	Short 3rd, 4th metatarsals	22.3	1.87	Normal stature	–2.56	Under Weight
BDA-10	S	8YM	Missing middle phalange of hands	Short feet	15.1	–3.45	Short stature	–1.99	Healthy Weight
BDA-11	S	10YM	Short hands	Short feet	12.7	–2.58	Short stature	–9.2	Under Weight
BDA-12	S	10YF	Short hands	Short feet	12.7	–3.08	Short stature	1.62	Healthy Weight
BDA-13	S	10YM	Short hands	Short feet	13.6	–2.72	Short stature	–1.11	Healthy Weight
BDA-14	S	11YM	Short hands	Short feet	15.7	–3.04	Short stature	–2.66	Under Weight

We compare the 14 BDA cases with age-sex matched 14 controls. The phenotype of BDA is shown in *figure 3*. Out of 14 Patients, 8 were females and 6 were male in both patients and controls shown in *figure 4*. By comparing the Mean ±SD of patients with controls, we found highly significant p-values in ratios of 2P:3P, 2P:4P, 2P:5P, 2L:3L, 2L:4L, 2L: 5L, 2D:1D. This predicted abnormal or short/missing proximal, middle, and distal phalanges of BDA patients. The third and fifth proximal phalanges are particularly short, the fourth proximal phalanges are long, the first distal phalanges are short, and the third, fourth, and fifth phalanges are long, the middle phalanges were found missing in familial case and short in other individuals. Graphical representation shown in supplementary figure 1S. Metacarpals were found to be unaffected. BDA in both right and left hands were found to have symmetrical involvement. A descriptive statistical comparison along with (unpaired t – test) of the left hand of BDA against controls of left hands is shown in table 2.

Table 2: Descriptive statistics showed significant p values in ratios of 2P:3P, 2P:4P, 2P:5P, 2L:3L, 2L:4L, 2L: 5L, 2D:1D This predicted short middle, proximal phalange of Brachydactyly A patient with respect to controls

Parameters	Brachydactyly A (BDA)				Control				
	Mean	Std. Deviation	95% CI Lower Bound	95% CI Lower Bound	Mean	Std. Deviatioin	95% CI Lower Bound	95% CI Upper Bound	Brachydactyly A (BDA) Vs Controls P value
2MC:1MC	1.470	0.257	1.322	1.620	1.48	0.218	1.362	1.608	0.6263
2MC:3MC	1.010	0.134	0.942	1.097	1.05	0.066	1.020	1.095	0.3457
2MC:4MC	1.150	0.169	1.054	1.249	1.18	0.115	1.119	1.252	0.7597
2MC:5MC	1.300	0.147	1.216	1.386	1.32	0.078	1.283	1.368	0.9909
2P:1P	1.540	0.427	1.300	1.794	1.29	0.109	1.240	1.357	0.3008
2P:3P	1.03	0.955	0.955	1.110	0.930	0.024	0.886	0.916	<0.0001****
2P:4P	0.237	0.059	0.202	0.271	0.968	0.050	0.940	1.001	<0.0001****
2P:5P	1.83	0.351	1.627	2.033	1.210	0.068	1.680	1.250	<0.0001****
2L:3L	0.821	0.089	0.791	0.947	0.845	0.093	0.801	0.910	0.0036**
2L:4L	0.838	0.112	0.762	0.913	0.898	0.086	0.862	0.960	0.0004*
2L:5L	0.991	0.183	0.868	1.115	1.26	0.151	1.990	1.374	0.0003***
2D:1D	1.040	0.251	0.900	1.191	0781	0.094	0.732	0.845	0.002***
2D:3D	0.959	0.083	0.911	1.008	0.985	0.065	0.956	1.029	0.1030
2D:4D	0.944	0.080	0.898	0.991	0.974	0.082	0.925	1.026	0.6755
2D:5D	1.000	0.155	0.915	1.094	1.300	0.838	0.816	1.851	0.1009

We enrolled 17 individuals with BDE based on phenotype and also evaluated the genotype by whole exome sequencing and Sanger sequencing. The phenotype of BDE patients with upper and lower limb manifestations, BMI, height for age, weight for age interpretation are showed in supplementary table 2S. In 17 BDE patients, we have 3 affected individuals of the same family, the rest belonged to unrelated families. The patients BMI, height for age and weight for age were predicted as short stature. Out of 17 patients, Out of 17 Patients, 11 were males and 6 were female in both patients and controls. 4 patients of BDE non-syndromic and syndromic patients were found with short stature out of which 2 were females and 2 were males. One male was noticed with tall stature showed in *figure 5*.

Table 2: Phenotypic appearance of BDE patients, interpret non- syndromic and syndromic (NS/S) patients, BMI, height for age, weight for Age

Patient ID's	NS/S	Age/ Gender	Upper limb	Lower Limb	BMI	Height for Age (HAZ)	Height	Weight for Age (WAZ)	Weight
BDE-1	NS	21YF	Short 3rd, 4th, 5th metacarpals	Short 3rd, 4th, 5th metatarsals	21.67	0.86	Tall stature	1.98	Healthy weight
BDE-2	NS	11YF	Short 3rd, 4th, 5th metacarpals	Short 3rd, 4th, 5th metatarsals	17.22	–448	Short stature	–2.86	Under weight
BDE-3	NS	11YF	Short 3rd, 4th, 5th metacarpals	–	18.37	–0.53	Normal stature	–1.5	Healthy weight
BDE-4	NS	43YF	Short 3rd, 4th, 5th metacarpals	Short 3rd, 4th, metatarsals	21.7	–4.6	Short stature	–4.97	Under weight
BDE-5	NS	11YF	Short 3rd, 4th, 5th metacarpals	–	16.66	–0.9	Normal stature	0.27	Healthy weight
BDE-6	NS	11YM	Short 3rd, 4th, 5th metacarpals	Short 3rd, 4th, 5th metatarsals	15.53	0.46	Normal stature	0.99	Healthy weight
BDE-7	S	17YM	Short 3rd, 4th, 5th metacarpals	Short 3rd, 4th, 5th metatarsals	17.33	0.86	Tall stature	–2.98	Under weight
BDE-8	S	10YF	4th metacarpal	Normal feet	14.24	–1.01	Normal stature	–4.71	Under weight
BDE-9	S	10YF	Short 3rd, 4th, 5th metacarpal	Normal feet	19	–0.52	Normal stature	–3.25	Under weight

Patient ID's	NS/S	Age/ Gender	Upper limb	Lower Limb	BMI	Height for Age (HAZ)	Height	Weight for Age (WAZ)	Weight
BDE-10	NS	11YF	3rd, metacarpal	Normal feet	14	0.39	Normal stature	–0.02	Healthy weight
BDE-11	NS	36YF	3rd, 4th, metacarpal	Normal feet	12.12	–0.04	Normal stature	–1.81	Healthy weight
BDE-12	NS	12YM	3rd, 4th, metacarpal	Normal feet	14.3	0.86	Normal stature	–0.17	Healthy weight
BDE-13	NS	31YF	3rd, 4th, metacarpal	Normal feet	23	–1.97	Normal stature	–2.32	Under weight
BDE-14	S	19YM	3rd, 4th, metacarpal	Normal feet	13.34	–5.16	Short stature	–5.26	Under weight
BDE-15	NS	13YF	3rd, 4th, metacarpal	Normal feet	16	0.87	Normal stature	–0.76	Healthy weight
BDE-16	NS	7YM	3rd, 4th, metacarpal	Normal feet	12	0.9	Normal stature	–0.09	Healthy weight
BDE-17	NS	12YM	3rd, 4th, metacarpal	Normal feet	13.6	–4.24	Short stature	–4.4	Under weight

We compared 17 BDE patients with 17 controls. By comparing the Mean ±SD of patients with controls, we found highly significant p values in ratios of 2MC:3MC, 2MC:4MC, 2MC:5MC, 2P:4P. Graph showed in the supplementary figure 1S. This predicted short 3rd, 4th, 5th metacarpals and 4th proximal phalange of BDE with respect to controls. Other phalanges were found to be unaffected. BDE affecting both right and left hands also showed symmetrical involvement. A descriptive statistical comparison of the left hand of BDE against controls of left hands is shown in *table* 3.

Table 3: Descriptive statistics showed significant p values in ratios of 2MC: 3MC, 2MC:4MC, 2MC:5MC, 2P:4P. This predicted short 3rd, 4th, 5th metacarpals and 4th proximal phalange of Brachydactyly E patients with respect to controls

Parameters	Brachydactyly A (BDA)				Control				
	Mean	Std. Deviation	95% CI Lower Bound	95% CI Lower Bound	Mean	Std. Deviatioin	95% CI Lower Bound	95% CI Upper Bound	Brachydactyly E (BDA) Vs Controls P value
2MC:1MC	1.490	0.090	1.446	1.538	1.498	0.198	1.397	1.599	0.7661
2MC:3MC	1.130	0.121	1.075	1.204	1.05	0.064	1.020	1.087	0.004**
2MC:4MC	1.480	0.275	1.341	1.624	1.18	0.112	1.127	1.243	0.0001***
2MC:5MC	1.600	0.318	1.438	1.243	1.31	0.078	1.280	1.359	0.0009***
2P:1P	1.270	0.091	1.226	1.316	1.32	0.106	1.265	1.375	0.1521***
2P:3P	0.898	0.086	0.857	0.938	0.903	0.024	0.892	0.918	0.6644
2P:4P	0.982	0.070	0.142	0.210	0.968	0.050	0.944	0.994	<0.0001****
2P:5P	1.150	0.162	1.072	1.237	1.22	0.075	1.186	1.263	0.5121
2L:3L	0.847	0.104	0.794	0.903	0.845	0.093	0.799	0.892	0.3655
2L:4L	0.878	0.128	0.813	0.943	0.898	0.086	0.854	0.940	0.8717
2L:5L	1.280	0.180	1.186	1.375	1.59	1.35	0.896	2.292	0.4801
2D:1D	0.944	0.718	0.640	0.840	0.740	0.019	0.790	1.458	0.7793
2D:3D	1.120	0.649	0.818	1.048	0.935	0.225	0.706	1.518	0.7667
2D:4D	1.110	0.790	1.458	0.727	0.924	0.223	0.706	1.518	0.8999
2D:5D	1.280	1.090	0.727	1.855	1.23	0.868	0.786	1.674	0.5231

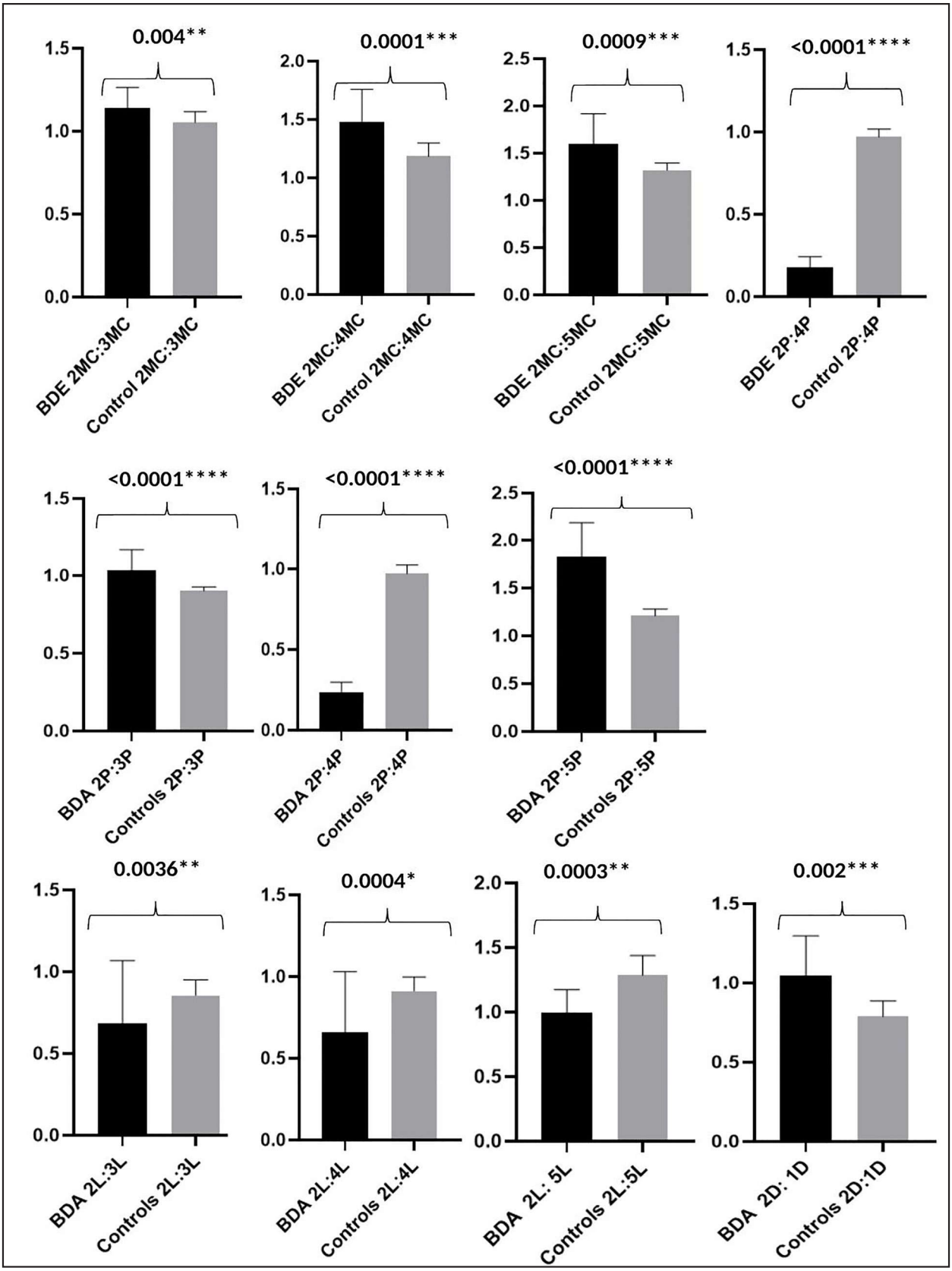

Figure 1: Graphical representation of BDA, BDE against control with significant p-value (X-axis represent: type of the phalanges and metacarpal, Y-axis represent: metacarpals and phalanges ratio of BDE, BDA and Controls)

DISCUSSION

Congenital autopod anomalies are very commonly found in the paediatric patients. The prevalence of the congenital anomalies of autopod is 1:800 living new-borns.[1,10] The congenital autopod anomalies are inherited in autosomal dominant pattern. The overall prevalence is 7.9/10,000 live births. Most are due to primary intrauterine growth inhibition, or disruptions secondary to intrauterine destruction of normal embryonic tissues. The upper extremities are more commonly affected. Anthropometry evaluation is concerned with quantitatively assessing the human body and skeleton and the correlations of metacarpals and phalanges of limb or hand proportions. No existing studies have described methodologies for predicting the metacarpal length and phalangeal length of affected brachydactyly patients. We have found that based on ratios of metacarpals 2MC: 3MC, 2MC:4MC, 2MC:5MC and 2P andproximal phalange to the other proximal phalanges, 2P: 1P, 2P:3P, 2P:4P, 2P:5P, 2L middle phalange to the other middle phalanges 2L:3L, 2L:4L, 2L:5L, 2D distal phalange to the other distal phalanges 2D:1D, 2D:3D, 2D:4D, 2D:5D can be compared for objective results. Evaluating measurements on X-ray films by anthropometric evaluation tools is a gold standard method to compare the growth variations in the human body and can ensure better comparisons. Its applications require normalized techniques and distinct milestones to guarantee that the information is reproducible, particularly for calculating different parameters in different parts of the skeleton.

CONCLUSIONS

We utilized a new way to analyse the types of brachydactyly: BDA and BDE by using ratios of the length of metacarpals and phalanges of the hands using X-ray films. We have demonstrated that this technique is highly reproducible and not very expensive. This can also be used in further studies for metacarpals and phalanges bone length assessment as a marker of other hand anomalies like asymmetric polydactyly and syndactyly. The limitation of this study is we are unable to recruited large sample size due to rarity of the BDA individual's cases and covid-19 pandemic reason.

Recommendations

- The anthropometric measurement tools may be used to evaluate X-rays from other skeletal parts of the body such as the foot.
- The use of other parameters in addition to hand length can be used for detailed evaluation of suspected skeletal dysplasia.
- This study is useful in updating skeletal dysplasia classification.

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REFERENCES

1. Jurcă M.C., Bembea M.A., Şandor M.I., Zaha D.C., Negrean R.A., Vesa C.M., Jurcă A.A., Moisa F.C., Vicaş L.G., Paul C., Cheregi S.D. Congenital anomalies of digits—A clinical-epidemiological study of 301 patients. Rom J Morphol Embryol. 2019 Jan 1; 60(4): 1221-6.
2. Hall C.M. International nosology and classification of constitutional disorders of bone (2001). American journal of medical genetics. 2002 Nov 15; 113(1): 65-77. https://doi.org/10.1002/ajmg.10828
3. Unger S., Ferreira C.R., Mortier G.R., *et al.* Nosology of genetic skeletal disorders: 2023 revision. American Journal of Medical Genetics Part A. 2023 May; 191(5): 1164-209. https://doi.org/10.1002/ajmg.a.63132
4. Pereda A., Garin I., Garcia-Barcina M., *et al.* Brachydactyly E: isolated or as a feature of a syndrome. Orphanet journal of rare diseases. 2013 Dec; 8: 1-4. https://doi.org/10.1186/1750-1172-8-141
5. Temtamy S.A., Aglan M.S. Brachydactyly. Orphanet journal of rare diseases. 2008 Dec;3:1-6. https://doi.org/10.1186/1750-1172-3-15
6. David A., Vincent M., Quéré M.P., Lefrançois T., Frampas E. Isolated and syndromic

brachydactylies: Diagnostic value of hand X-rays. Diagnostic and Interventional Imaging. 2015 May 1; 96(5): 443-8. https://doi.org/10.1016/j.diii.2014.12.007

7. Leung A.O., Poon A.C., Wang X., *et al*. Suppression of apoptosis impairs phalangeal joint formation in the pathogenesis of brachydactyly type A1. Nature Communications. 2024 Mar 12;15(1):2229. https://doi.org/10.1038/s41467-024-45053-0

8. Sun J., Yang N., Xu Z., Cheng H., Zhang X. A novel heterozygous mutation in PTHLH causing autosomal dominant brachydactyly type E complicated with short stature. Molecular Genetics & Genomic Medicine. 2024 Feb; 12(2): e2393.https://doi.org/10.1002/mgg3.2393

9. Zaib T, Rashid H, Khan H, Zhou X, Sun P. Recent advances in syndactyly: Basis, current status and future perspectives. Genes. 2022 Apr 27; 13(5): 771. https://doi.org/10.3390/genes13050771

10. Ahmad Z, Liaqat R, Palander O, *et al*. Genetic overview of postaxial polydactyly: updated classification. Clinical Genetics. 2023 Jan;103(1): 3-15. https://doi.org/10.1111/cge.14224

11. Manning J.T., Trivers I.V. R.L., Thornhill R., Singh D. The 2nd: 4th digit ratio and asymmetry of hand performance in Jamaican children. Laterality: Asymmetries of Body, Brain and Cognition. 2000 Apr 1; 5(2): 121-32. https://doi.org/10.1080/00222937100770081

12. Sephien A., Bethel C.F., Doyle C.M., Gulick D., Smith C.J., Schwartz-Fernandes FA. Morphometric analysis of the second through fifth metacarpal through posteroanterior X-Rays. Clinical Anatomy. 2020 Oct; 33(7): 1014-8. https://doi.org/10.1002/ca.23528

13. Foley D.P., Cox C.T., Foley A.S., *et al*. Predicting metacarpal length using paired ratios with bilateral X-ray films. SAGE Open Medicine. 2021 Dec; 9 https://doi.org/10.1177/20503121211064177