PHENOTYPIC SPECTRUM AND CYTOGENETIC EVALUATION OF POLYDACTYLY: A CLINICO-GENETIC STUDY

Dr.Bindurani M K* [1], Dr.Asha K R [2], Dr.Meenakshi Bhatt [3], Dr.jayaram S Kadandale[4], Dr.Lakshmi Prabha subhash [5], Mrs. Harshal [6]

[1]Professor,Dept. of Anatomy, Sri Siddhartha medical college, Tumkur. Karnataka.India. 572107
Contact no : 9880041364
Drbindumk3@gmail.com
[2]Prof & Head, Dept. of Anatomy, Siddaganga medical college & research institute, Tumkur. Karnataka. 572107
Contact no : 9886051964
drashakeshavraj@gmail.com
[3]Clinical Geneticist, Sri Siddhartha Medical College, Tumkur, Karnataka- 572107
Bhat.meena@gmail.com
[4]Consultant Geneticist, Sri Siddhartha Medical College, Tumkur, Karnataka- 572107
jayaram@chg.res.in
[5]Prof & Head, Dept. of Anatomy, Sri Siddhartha institute of health sciences, T Begur. Karnataka.
[6]Research Associate, Sri Siddhartha Medical College, Tumkur, Karnataka- 572107
Harshal.vish@gmail.com

Corresponding Author : Dr.Bindurani M K
Email id : Drbindumk3@gmail.com

ABSTRACT

Background: Polydactyly is a congenital anomaly characterized by the presence of an extra digit in the hand or foot. It may occur as an isolated malformation or as part of a genetic syndrome. The reported incidence ranges from 0.37 to 1.2 per 1000 live births, varying among different populations. Based on the location of the extra digit, polydactyly is classified as postaxial, preaxial, or central. Limited descriptive data are available on the phenotypic and genetic characteristics of polydactyly in Asian populations. The present study aimed to evaluate the phenotypic manifestations and chromosomal findings in subjects with polydactyly.

Methods: A total of 105 subjects with polydactyly who attended the Genetics Laboratory were included in the study. Clinical evaluation, family history, assessment of parental consanguinity, and chromosomal analysis were performed.

Results: Of the 105 cases, 43 (41%) were classified as syndromic polydactyly due to the presence of associated systemic malformations, while 62 (59%) were non-syndromic. Sporadic occurrence was more common in syndromic cases (72.1%), whereas familial occurrence was observed in nearly half of the non-syndromic cases (48.4%). Parental consanguinity was present in 51 cases, including 28 syndromic and 23 non-syndromic subjects. In both groups, involvement of the hands was more frequent than that of the feet. Postaxial polydactyly was the predominant phenotype. The most common associated systemic manifestations were obesity and ocular abnormalities (17%), followed by craniofacial and oral anomalies (12%), developmental delay (10%), cardiovascular anomalies (3%), and genitourinary anomalies (3%). Chromosomal analysis revealed a normal karyotype in the majority of cases. Abnormalities were detected in five subjects, including four cases of trisomy 21 and one non-syndromic subject with a pericentric inversion of chromosome 7 [46,XY,inv(7)(p15q11.23)].

Conclusion: Overall, the findings underscore the importance of comprehensive clinical evaluation, detailed family history, and appropriate genetic investigations in all cases of polydactyly. Early identification of associated anomalies enables timely intervention, multidisciplinary management, and effective genetic counselling, thereby improving patient care and prognostic assessment.

Key words: Polydactyly, Syndromic polydactyly, Congenital anomalies, systemic malformations, Chromosomal abnormalities.

How to cite this article: Bindurani MK, Asha KR, Bhatt M, Kadandale JS, Subhash LP, Harshal. Phenotypic Spectrum and Cytogenetic Evaluation of Polydactyly: A Clinico-Genetic Study. Int J Drug Deliv Technol. 2026;16(71s): 954-959. DOI: 10.25258/ijddt.16.71s.111

INTRODUCTION

The term polydactyly is characterized by the presence of an additional digit either in finger or toe. The extra digit may be fully or partially developed with or without bone. It can also be called polydactylia, polydactilism or hyperdactyly.

Polydactyly can occur as an isolated malformation or as part of a genetic syndrome. It is a common limb related birth defect with an incidence between

0.37 and 1.2 per 1000 live births, depending on race.[1] Polydactyly can be postaxial (on the ulnar or fibular side of the limb), preaxial (on the radial or tibial side of the limb) or midline. and can be unilateral or bilateral. The occurrence of postaxial polydactyly in the general population varies among different racial groups and is 10 times more frequent in Blacks than in Caucasians [2]. In terms of clinical variability, polydactyly is one of the most heterogeneous types of digit anomaly[3]

About 15% of children born with polydactyly have other congenital anomalies, usually as part of a defined syndrome like familial polydactyly (inherited), Ellis- van Creveld Syndrome, Carpenter Syndrome, Trisomy 13, Rubenstein-Taybi Syndrome, Smith-Lemli-Opitz, Laurence-Moon-Biedl Syndrome, and Asphyxiating Thoracic Dystrophy etc.[5] There are relatively less descriptive data on polydactyly for Asian populations in the medical literature [4]. The present study aims to study phenotypic manifestations & chromosomal analysis of polydactyly subjects.

METHODOLOGY:

It was a descriptive study conducted in the genetic laboratory, department of Anatomy, Sri Siddhartha Medical College, Tumkur, Karnataka, India. A total of 105 polydactyly subjects who attended Genetic lab were included in the study. Institutional Ethics Clearance was taken. Written consent was obtained from all the subjects/parents (in children) after explanation of the nature and possible consequences of the study. In each case family history, history of consanguinity and head to toe examination was done. In all the cases of polydactyly, three generation pedigree charts and associated clinical features were observed.

The chromosomal studies from peripheral blood were carried out.

.

RESULTS:

Of the total 105 cases studied, 43 subjects presented with associated systemic malformations and were classified as **syndromic polydactyly**, while the remaining 62 cases were categorized as **non-syndromic/isolated polydactyly**.

Among the 43 syndromic cases, 22 (51.2%) were males and 21 (48.8%) were females. In the non-syndromic group, 33 (53.2%) were males and 29 (46.8%) were females.

With respect to inheritance pattern, sporadic occurrence was observed in 31 (72.1%) of the syndromic cases, whereas 12 (27.9%) had a positive familial history. In contrast, among the non-syndromic cases, 32 (51.6%) were sporadic and 30 (48.4%) showed familial occurrence. occurrence pattern showed that syndromic polydactyly was predominantly sporadic, observed in 31 (72.1%) cases, with only 12 (27.9%) demonstrating familial occurrence.

Table 1: Distribution of polydactyly subjects in sporadic & familial occurrence

	Syndromic polydactyly(43)		Non Syndromic polydactyly(62)	
	Sporadic occurence	Familial occurence	Sporadic occurence	Familial occurence
Males	16	6	13	20
Females	15	6	19	10
	31	12	32	30
	43		62	

Out of 105 cases, 51 cases were associated with parental consanguinity out of which 28 were syndromic & 23 were non syndromic. (Table 2)

Table 2 – Proportion of Consanguinity in polydactyly subjects

Consanguinous				**Non consanguineous**			
Syndromic		Non syndromic		Syndromic		Non syndromic	
Sporadic occurence	Familial occurence	Sporadic occurence	Familial occurence	Sporadic occurence	Familial occurence	Sporadic occurence	Familial occurence
20	8	14	9	11	4	18	21
28		23		15		39	

Table 3 : Involvement of autopods with type of polydactyly in syndromic polydactyly subjects
H-hands, F --Foot

	Syndromic sporadic(31)				Syndromic familial(12)			
	Hands only	Foot only	Both H& F		Hands only	Foot only	Both H &F	
Pre axial	5	2	6	13	2	0	0	2
Post axial	9	1	8	18	6	2	2	10
	14	3	14		8	2	2	

	Syndromic sporadic(31)			**Syndromic familial(12)**			
	H	F	H& F	H	F	H &F	
Pre axial	**5**	**2**	**6**	**2**	**0**	**0**	**15**
Post axial	**9**	**1**	**8**	**6**	**2**	**2**	**28**
	14	**3**	**14**	**8**	**2**	**2**	

Table 4 : Involvement of autopods with type of polydactyly in Non syndromic polydactyly subjects

	Non Syndromic sporadic(32)				NonSyndromic familial(30)			
	Hands only	Foot only	Both H& F		Hands only	Foot only	Both H& F	
Pre axial	9	0	0	9	6	2	1	9
Post axial	10	5	8	23	9	5	7	21
	19	5	8		15	7	8	

In both syndromic & non syndromic polydactyly cases hands were more commonly involved than feet in all categories. Overall, post-axial polydactyly was the predominant phenotype in both syndromic and non-syndromic groups, regardless of

inheritance pattern, with isolated hand involvement being the most frequent anatomical presentation.

Table 5: Association of systemic malformations in polydactyly subjects

	Males		Female		
System	sporadic	familial	Sporadic	Familial	**Total**
Obesity	7	1	6	3	17 (16.19%)
Ocular	7	1	6	3	17 (16.19%)
Oro facial	2	3	6	1	12 (11.4%)
Developmental delay	3	2	4	1	10 (9.5%)
CVS		2	1	0	3 (2.8)
Genito urinary	3	0	0	0	3 (2.8%)
Clubbed foot	0	0	1	0	1(0.9%)
Multiple lipoma	1	0	0		1 (0.9%)
Respiratory system	1	0	0		1 (0.9%)
syndactyly	0	2	0	1	3

The most common systemic manifestations observed were obesity and ocular abnormalities which included congenital cataract, retinitis pigmentosa ,progressive night blindness and glaucoma. The Oro facial features identified included micrognathia, cleft lip, macroglossia, a flat nasal bridge, dental abnormalities and hypertelorism. Cardiovascular abnormalities comprised congenital heart diseses. Genito urinary anomalies included hypogonadism, bilateral undescended testes (cryptorchidism) & poly cystic kidney disease. In the present study, the majority of polydactyly cases showed a normal chromosomal pattern , while a small proportion (5 cases) demonstrated abnormalities, which included 4cases of trisomy 21 & one non syndromic subject had pericentric inversion of chromosome 7. 46,XY,inv(7)(pq15q11.23)**Table 6. Chromosomal studies in polydactyly**

	Normal	**Abnormal**
Chromosomal study	100	5 (4 cases of trisomy 21 1 case of pericentric inversion of chromosome 7 46,XY,inv(7)(pq15q11.23)

DISCUSSION :

Of all congenital hand malformations, polydactyly is the most frequently observed anomaly at birth and it is even one of the most frequent congenital disorders in general. It has been shown that there are several routes to developmental perturbation during digital morphogenesis that may lead to additional digits. [6]

Polydactyly results from defective patterning of the anterior-posterior axis of the developing limb .[7]

Syndromic Polydactyly occurred sporadically in 72.1% of cases where as non syndromic polydactyly had balanced proportion in sporadic & familial occurrence. Earlier study on non syndromic polydactyly subjects in Chinese population showed that it occurred sporadically in 95.2% of the subjects. [8]

In an another study, 64.5% of the subjects had a sporadic occurrence and 35.5% were familial.[4]

Hand involvement was observed more commonly than foot involvement in both syndromic and non-syndromic polydactyly cases. Similar findings were noted in previous studies.(4,8). Ying Xiang, Jingxia Bian, Zhigang Wan, Yunlan X u, Qihua Fu. Clinical study of 459 polydactyly cases in China, 2010 to 2014. Congenital Anomalies 2016; 56, 226–232)

Postaxial and preaxial types affected the upper and lower limbs and the right and left limbs at different rates in different geographical population. Post-axial polydactyly was more frequently observed than pre-axial involvement in both syndromic & non syndromic groups. This finding is consistent with existing literature, which identifies post-axial polydactyly as the most frequent form, particularly in isolated (non-syndromic) cases. [1]. One of the study done in America shows that incidence of post axial polydactyly as highest and hands were involved commonly [8]. In a study done in Pakistan, post axial type was most common.[4]. In contrast, in the recruited Chinese cohort, preaxial polydactyly type I was found to be the most common and postaxial polydactyly type A was the second most [9].

In a study on neonates, the most frequently observed forms of polydactyly were preaxial hand polydactyly and postaxial foot polydactyly. [10] Lee Y, Gang M, Chang M, Song W, Kim S. Clinical study of 31 polydactyly cases admitted to neonatal intensive care unit. *Perinatology*. 2019;30(3):147-153. doi:10.14734/PN.2019.30.3.147) Sex predilection was observed in previous studies. Males were affected more compared to females.[4]. But our study does not show any such significant sex predilection.

The present study demonstrates that polydactyly is frequently associated with systemic malformations, with obesity and ocular anomalies being the most common. These findings are consistent with previous reports highlighting the association of polydactyly with multisystem disorders, particularly ciliopathies such as Bardet–Biedl syndrome, where retinal abnormalities and obesity are prominent features. [11] . The second most common congenital anomalies associated with polydactyly in our study is anomalies of face and oral cavity.[12] Franco B, Thauvin-Robinet C. Update on oral-facial-digital syndromes (OFDS). Cilia. 2016 May 2;5:12. doi: 10.1186/s13630-016-0034-4. PMID: 27141300; PMCID: PMC4852435. In previous study on neonates the most frequent congenital anomalies noted were Cardiac anomalies followed by (35%) genitourinary tract anomalies (22%), central nervous system anomalies (22%) & gastrointestinal tract anomalies (19%)

Overall, these findings underscore the importance of comprehensive systemic evaluation in individuals with polydactyly to facilitate early diagnosis of associated anomalies and enable appropriate multidisciplinary management and genetic counseling.

Chromosomal analysis revealed abnormalities in 5 cases out of which 4 were trisomy 21. Earlier studies have reported the assisciation of polydactyly with trisomy 21.[13.14]

In one non syndromic polydactyly patient there was pericentric inversion of chromosome 7. Similar chromosomal abnormality was reported in a child with Hand foot genital syndrome.[15]

Overall, these findings highlight the importance of chromosomal analysis in selected cases, particularly when polydactyly is associated with other congenital anomalies, to aid in accurate diagnosis, prognosis, and genetic counseling.

CONCLUSION:

Overall, the findings underscore the importance of comprehensive clinical evaluation, detailed family history, and appropriate genetic investigations in all cases of polydactyly. Early identification of associated anomalies enables timely intervention, multidisciplinary management, and effective genetic counselling, thereby improving patient care and prognostic assessment.

REFERENCES:

1. Castilla EE, Paz JE, Mutchinick O, Muñoz E, Giorgiutti E, Gelman Z. Polydactyly: A genetic study in South America. **Am J Hum Genet.** 1973;25:405-412.
2. Radhakrishna U, Blouin JL, Mehenni H, Patel UC, et al. Mapping one form of autosomal dominant postaxial polydactyly type A to chromosome 7p15-q11.23. **Am J Hum Genet.** 1997;61:597-604.
3. Temtamy SA, McKusick VA. **The genetics of hand malformations.** Birth Defects Orig Artic Ser. 1978;14(3):1-619.
4. Malik S, Saifullah, Afzal M, Lal K, Haque S. Clinical and descriptive genetic study of polydactyly: A Pakistani experience of 313 cases. **Clin Genet.** 2014;85(5):482-486.
5. Milton H. Polydactyly and syndactyly. **Hershey Medical Center.** 2010.
6. Talamillo A, Bastida MF, Fernandez-Teran M, Ros MA. The developing limb and the control of the number of digits. **Clin Genet.** 2005;67:143-153.
7. Biesecker LG. Polydactyly: how many disorders and how many genes? 2010 update. Dev Dyn. 2011 May;240(5):931-42. doi: 10.1002/dvdy.22609. Epub 2011 Mar 28. PMID: 21445961; PMCID: PMC3088011.)
8. Woolf CM, Myrianthopoulos NC. Polydactyly in American Negroes and Whites. **Am J Hum Genet.** 1973;25:397-404.
9. Xiang Y, Bian J, et al. Clinical study of 459 polydactyly cases in China, 2010 to 2014. Congenit Anom (Kyoto). 2016;56:226-232.
10. . Lee Y, Gang M, Chang M, Song W, Kim S. Clinical study of 31 polydactyly cases admitted to neonatal intensive care unit. *Perinatology*. 2019;30(3):147-153. doi:10.14734/PN.2019.30.3.147
11. Forsythe E, Beales PL. Bardet-Biedl syndrome. **Eur J Hum Genet.** 2013;21(1):8-13.

12. Franco B, Thauvin-Robinet C. Update on oral-facial-digital syndromes (OFDS). Cilia. 2016 May 2;5:12. doi: 10.1186/s13630-016-0034-4. PMID: 27141300; PMCID: PMC4852435

13. Castilla EE, Lugarinho R, da Graça Dutra M, Salgado LJ. Associated anomalies in individuals with polydactyly. Am J Med Genet. 1998 Dec 28;80(5):459-65. doi: 10.1002/(sici)1096-8628(19981228)80:5<459::aid-ajmg5>3.0.co;2-g. PMID: 9880209.

14. Barnes CJ, Taqi M, De Cicco FL. Supernumerary Digit. StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK564405
15. .
16. Watson CM, Crinnion LA, Harrison SM, et al. A chromosome 7 pericentric inversion defined at single-nucleotide resolution using diagnostic whole genome sequencing in a patient with hand-foot-genital syndrome. **PLoS One.** 2016;11(6):e0157075.