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A Case Report of Genetic Malformations in a Jatapu Tribal Girl of Andhra Pradesh

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KEY WORDS Genetic malformations; polydactyly; brachydactyly

ABSTRACT The combiantional occurrence of congenital malformations, which are autosomal dominant viz., Polydactyly and Brachydactyly is evidient in an 8 year old girl child in a family belonging to a tribal group - the Jatapu of Vizianagaram District of Andhra Pradesh. The pedigree analysis reveals that the affected girl's parents and brother are normal. However, her parents are consanguineous by marriage showing first cross cousin type. The anthropometric data on hand yield low values compared to parents and sibs. The probabale reason for such rare combination of malformations together in an individual may be due to varied biochemical mechanism that underlie the manifestation of gene for incomplete penetrance or expressivity. The present study suggest further probe to ascertain linkage analysis at protein level.

INTRODUCTION

In man, several characters were observed controlled by either single gene or many genes. Among the single gene, both autosomal and sex-linked traits show dominance and recessive expressivity. The incidence of genetic malformations, though sporadic, gained prominence in human genetics since the pedigrees of the affected provide baseline information in turn gene structure and genetic code for practical application of results at species level. Population genetic surveys on different ethnic groups throw light on such rare genetic disorders. Farabee (1905) first described in his paper on 'Inheritance of Digital Malformations in Man' a pedigree showing autosomal dominant character viz., Brachyphalangy later referred as Brachydactyly or Short-Fingeredness. Mohr and Wriedt (1919) reported a child with multiple malformations born (but died) to the consanguineous parents who were moderate brachydactyly bearers. Likewise, scholars ascertained other genetic abnormalities (Curtstern 1961; Garrod 1902; McKusick 1983).

The present paper reports, for the first time, a combination of autosumal dominant characters-Polydactyly and Brachydactyly besides other abnormalities in a tribal family-the Jatapu, from Vizianagaram District of Andhra Pradesh, India.

The Jatapu is a Scheduled Tribe of Andhra Pradesh inhabiting coastal districts. They speak 'kuvvi' dialect which identifies them with the Khonds (Rao 1985). Their economy is Agro-Forest based. They practice shifting cultivation or 'Konda Podu' and terrace cultivation. They collect minor forest produce to supplement their economy. They prefer and practice consanguineous marriages accounting 59.50% with an average Inbreeding coefficient of 0.0372 for Autosumal genes and 0.0256 for sex-linked genes (Durga Gandhi 1998).

MATERIALS AND METHODS

Nutritional Anthropometric study was conducted among the Jatapu children during December, 1997 in ten (10) villages viz., Mettu Valasa, Kotha Valasa, Boorji Valasa, Sompigaun, Dalai Valasa, Diguwa Kasai Valasa, Mavudi, Mavudi Kotha Valasa, Anti Valasa and Loya Valasa of Salur Mandal of Vizianagaram District, Andhra Pradesh (India). In Mavudi Village, a Jatapu girl, aged 8 years, was found with rare combination of genetic malformations i.e., extra-fingeredness or Polydactyly and short-fingeredness or Brachydactyly. A pedigree was constructed and examined her parents and sibs for such malformations. The type of consanguineity was also determined. Appropriate anthropometric measurements were taken on the girl besides observing the dentition, feet etc. Photographs were taken for effective visual presentations of malformations.

RESULTS

From the Pedigree (Fig.1) it is evident that only on girl child affected belonging to 3rd birth

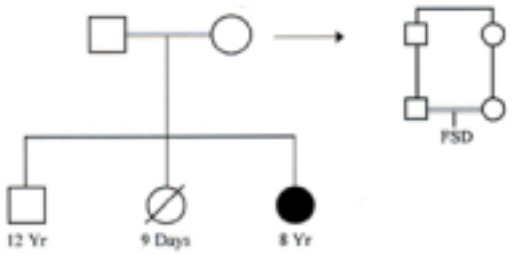

Fig 1. Pedigree showing genetic malformation

Table 1: Anthropometric data

Measurement (cm)	*R.Hand*	*L.Hand*	*6th Finger*	
			R	*L*
Lenth	9.50	9.80	1.90	2.00
Breadth	6.80	7.10	1.00	1.00
Index	71.58	72.45	52.63	50.00

Fig. 2. Hands showing both polydactyly and brachybactyly

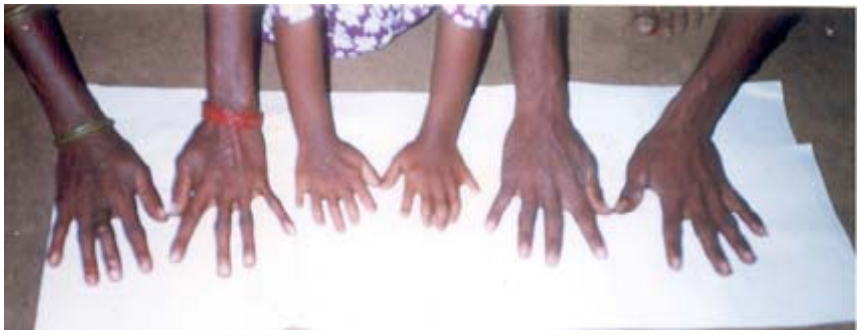

Fig. 3. Affected child with normal parents (hands)

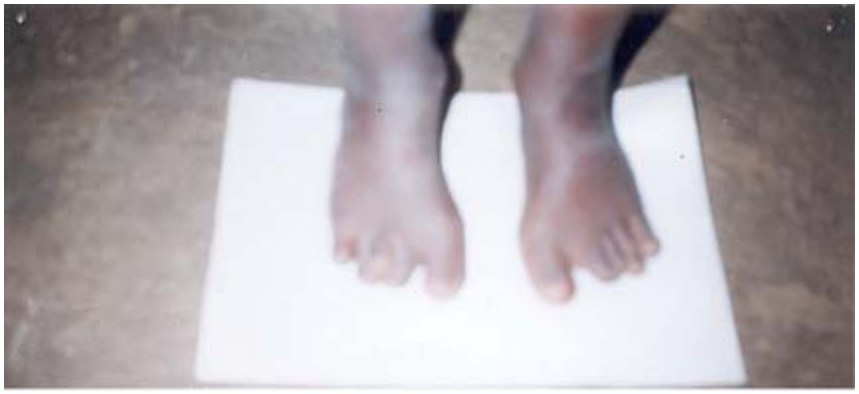

Fig. 4. Affected child feet showing malformations

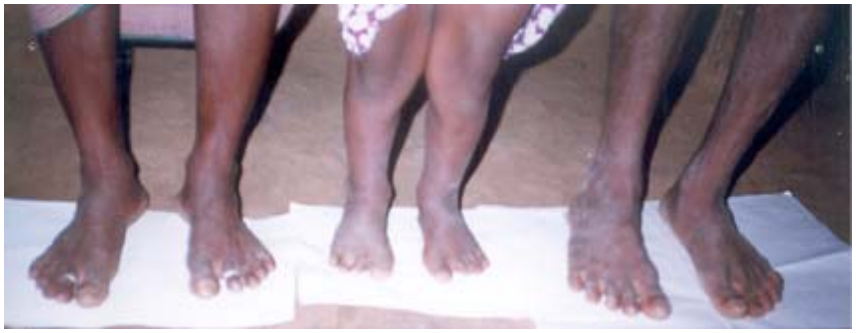

Fig. 5. Affected child with normal parents (feet)

Fig. 6. Mother and Father with affected daughter

order, Her brother aged 12 years is normal, while her sister died at the age of 9 days old with unknown reasons. Surprisingly, none of her parents show any evidence for any sort of malformations. Yet the parents are consanguineous of first cross-cousin that too Father Sister's Daughter (FSD) type. In general, the FSD occurs in this population with a frequency of 39% followed by 20.5% of MBD (Durga Gandhi 1998).

The anthropometric data (Table 1), and Photographs (Fig. 2-6) reveal that the affected individual shows conspicuously the combination of malformations on both the hands with little bilaterality. The length and breadth of right hand is 9.5 cm and 6.8 cm while it is 9.8 cm and 7.1 cm for the left. The lenth and breadth of extra finger (6th) is 1.9cm and 1.0cm on the right and 2.0cm and 1.0cm on the left. Accordingly the indices vary. Regarding the feet they show short toes with gradual increase of curvature from great to little toe. In dentition, an incomplete development of Premolar in the maxilla and secluded incisors, besides lack of molar teeth in the mandible. The photographs provided exhibit certain observations on physical growth and development, body features etc.

DISCUSSION

Autosomal dominant characters arise due to mutations affecting structural protiens (McKusick 1983), They are independent of sex in transmission. In the present study, since none of the parents show any sort of malformation it may be viewed that either they lack the mutant gene or varied biochemical mechanism that underlie the manifestation of gene for incomplete penetrace or expressivity. Moreover, combinational occurence of two autosomal dominant characters in the same individual generate interest to probe further for linkage analysis at protein level.

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