The Journal of
Maharashtra Orthopaedic Association
March - 2006

The Mode of Inheritance and Other Variables in Idiopathic Talipes Equinovarus

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❖ Abstract ❖

This study is based on 558 unrelated families each having at least one child under treatment for talipes equinovarus in Govt. Hospital Kudal Oras.

A substantial familial correlation was established: first degree relatives (siblings and parents) had a 3.8 per cent occurrence rate about twenty one times as high as the normal rate, among second degree relative (aunts and uncles) a 0.9 per cent rate was established, about eight times the population rate and among third degree relative (cousins) a 0.2 per cent occurrence which is near to the incidence in the general population.

In the group with the positive family history 228 of 1410 siblings (16.4%) had talipes equinovarus. In the group of families with no other affected relative 340 of 12216 siblings (2.7%) were affected The statistical difference between these recurrence risks is highly significant ($p<0.005$).

There were 126(55.2%) affected males and 102(44.8%) affected females in the group having other affected relatives. In the group with a negative family history 172(50.1%) of the affected children were male and 168(49.9%) were female. This sex ratio of 1.2:1 is significantly different from the expected 2:1 but does differ from the 1:1 ratio observed in the normal population.

In the absence of a positive family history the risk of the deformity recurring in a subsequent sibling of the probands is no greater than in the general population. Whereas in the presence of a positive family history there is roughly a 20% chance of the deformity reappearing in additional children born to same parents.

Introdction

Congenital club foot is one of the most common birth defects, with a reported incidence of 2.45 per 1000 live births in Sindhudurg District.[1] The literature regarding the inheritance patterns in club foot is confusing, however as club foot may be caused by both environmental and genetic factors.

The most common form of congenital club foot is the idiopathic variety, found in the otherwise normal infant in whom no syndrome, Mendelien disorder, cytogenetic abnormality or extrinsic cause can be found. It should be a diagnosis of exclusion.

Palmer originally favored the theory of an autosomal dominant gene reduced penetrance as the cause, however with further data he altered his view to support a multifactorial system of inheritance possibly with intrauterine factors having some effect.[2]

Deformities which appear to be passed from parents to children have created special curiosity. Club foot has been recognized as one such condition. In his Jacksonian Prizc Essay of 1864, Adams reported two families with multiple occurrences of talipes equinovarus. The search for a hereditary pattern has been sporadic since that time and no definite conclusions have been reached.[3]

Material & Methods

This study is based on 558 unrelated families each having at least one child under treatment for talipes equinovarus in the Govt. Hospital Kudal Oras.

This hospital serves the greater part of the largely rural county of Sindhudurg District whose population is fairly stable so that it is relatively easy to trace families and it is possible to follow progress over long periods

All probands (index cases) and their siblings were examined and their parents interviewed by me. In 558 families there were 13626 children, including 1st, 2nd, 3rd, degree relatives, 568 of whom had talipes equinovarus. 228 (40.1%) of the families had one or more similarly affected relatives. In this group there were 46 (20.2%) families with affected mothers, 14 (6.1%) with affected fathers and 3 (1.3%) in which both parents were affected. Not a single even distant relative was affected in 330 (58%) families, now considered as a group with negative family history.

Refer Table - I

The number of club footed children born to mothers of various ages were obtained for each group.

The ratio of males to females with talipes equinovarus was obtained into the group with both positive and negative family histories.

For the purpose of analysis our data were divided into four groups.

Group 1 : The families with a negative history.

Group 2 : The families in which one parent with or without another relative was affected;

Group 3 : The families in which some relative other than a parent or sibling was affected and

Group 4 : The families about which we had complete information and which were related to the families under study.

To check the validity of these divisions a test for homogenecity was made among the three groups with a positive family history (Groups 2,3 and 4). The homogenecity of this group was then tested with the group against a negative family history as a final check.

Results

A substantial familial correlation was established. First degree relatives (siblings and parents) had a 3.8 per cent occurrence rate, about twenty one times as high as the normal rate, among second degree relative (aunts and uncles), a 0.9 per

Table - I

Talipes Equinovarus First Degree Relatives With The Same Depormity

	Father	Mother	Father and Mother both	Brothers	Sisters
Male index - 126	8 of 126	25 of 126	2 of 126	72 of 362	54 of 424
Female index - 102	6 of 102	21 of 102	1 of 102	58 of 232	44 of 392
Total	14 of 228	46 of 228	3 of 228	130 of 594	98 of 816
228 of 1410 siblings affected (16.4%)					

Table - II

Recurrence Risks For Talipes Equinovarus

	Frequency	Recurrence Risk (percent)
** Occurrence (high limit)	1:1000	0.10
** Occurrence (low limit)	1:1400	0.07
* Negative family history	340:12216	2.7
* Positive family history	228:1410	16.4
$X^2 = 7.5088$	$P < .005$	

* Figures for the positive and negative family histories are from the current series.

** Figures from Palmer R.M.

cent rate was established, about eight times the population rate and among third degree relative (cousins) a 0.2 per cent occurrence which is near to the incidence in the general population.

In the group with the positive family history 228 of 1410 siblings (16.4%) had talipes equinovarus. In the group of families with no other affected relative 340 of 12216 siblings were affected (2.7%) The statistical difference between these recurrence risks is highly significant ($p < 0.005$).

Refer Table - II

There were 126 (55.2%) affected males and 102 (44.8%) affected females in the group having other affected relatives. The sex ratio is 1.1:1 which differs significantly from the 2:1 ratio, suggested by previous workers at the 95 percent coufidence limit (.05 > .025). In the group with a negative family history 172 (50.1%) of the affected children were male and 168 (49.9%) were female. This sex ratio of 1.2:1 is significantly different from the expected

Table - III

Sex Ratios of Positive and Negative Family History Groups and Their Confidence Limits

	Male %	Female %	Sex Ratio	X^2 Test			
				1:1 Ratio		2:1 Ratio	
				X^2	P	X^2	P
Positie family history	55.2	44.8	1.2:1	0.1696	0.7 > p > 0.5	4.6660	0.5 > p > 0.025
Negative family history	50.1	49.9	1:1	5.2676	1 > 0.005	0.2761	0.75 > p > 0.50

Table IV

Talipes Equinovarus Ages of Mothers

	Age of Mother (Years)			
	15-24	25-29	30-24	35 & Over
* Observed Positive family history	86	94	38	10
* Observed Negative family history	92	176	58	14
** Expected	31.54	30.64	19.01	12.82
X 2/(3)= 1.48 p > 0.10 Not Significant				

* Figures for the positive and negative family histories are from the current series.

** Figures from Devon births central figures.

Table V

Talipes Equinovarus Birth Order

	Birth Order			
	1	2	3	4
* Observed Positive family history	136	56	24	12
* Observed Negative family history	168	118	25	29
** Expected	37.19	28.75	14.28	13.78
X 2/(3) = 4.97 p>0.10 Not Significant				

* Figures for the positive and negative family histories are from the current series.

** Figures from Devon birth national figures.

2:1 but does differ from the 1:1 ratio observed in the normal population.

Refer Table - III

There is a significant excess among young mother 78.5% (180/228) in both positive history and 78.8% (268/340) in negative history. Similarly significant excess seen among first born children with positive family history in 60.4% (136/228) and with negative family history 60.4% (168/340), statistically both not significant.

Discussion

Rather the authors felt that the club foot seen with congenital anomalies was secondary to the decreased capacity of the uterus resulting from early rupture of the amnion with the chorion remaining intact.[4] Torpin substantiated the role of the ammion in the production of congenital anomalies in numerous publications.

Clubbing of the feet can be produced experimentally in rats removing the aminiotic fluid from the amniotic sac during gestation.[5] Poswillo demonstrated defects in nine of forty-nine rat embryos following perforation of the amnion.

When certain data, found in virtually all published series are reviewed in summation, they seem to indicate that club foot is twice as common in males as in females contrary to this study observations that roughly half of the cases are bilateral and that the remaining cases are rather evenly divided between the right and left side.[6] Various authors have noted the incidence of positive family histories in their series.[7]

Table VI

Estimates of Positive Family History In Club Foot

Author	Percent of cases
Scaglietti	5.12
Seli	11.43
Kite	44.64
Macewen and associates	18.79
Ducci and Grilli	5.07
Fetscher	13.60
Isigkeit	4.95
Nilsonnc	12.50
Miiller	15.00
Panum	25.00
Ehreufried	50.00
R.S. Kulkarni (Prescent series)	**40.90**

The analysis of these data provides evidence that primary talipes equinovarus should not be considered a single entity; from present observations it should be divided into at least two groups. The * first of these may be partially or totally nonhereditary. It has a negative family history. It has a predominance of younger mothers. Its recurrence risk is not significantly higher than that of the general population The second group should be considered hereditary. Its recurrence risk is almost 20 percent.

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