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Original Article

Chromosomes, Dermatoglyphics and Polycystic Ovary Syndrome (pcos)

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ABSTRACT

Aims: To study and co-relate chromosomal abnormalities and dermatoglyphics in infertile female patients with PCOS. Method: 16 cases of PCOS and 16 normal females as controls were selected. Chromosomal study was done using whole blood culture method and GTG Banding in genetic laboratory. Palmar and Plantar Dermatoglyphics were studied using "Roller ink method" using Kore's duplicating ink and analyzed using Henry's system. Result: Incidence of chromosomal abnormalities is 81.25% (13/ 16 cases). Distribution of chromosomal abnormalities is as follows.

- Numerical Chromosomal abnormality (47,XXY Female)- 06.25%
- Autosomal translocation [t(14;7)(q21→31;q36)]- 06.25%
- Microdeletions like, (2)(q12) homo - 50%, (2)(q12) hetero - 25%, (10)(q25) hetero - 12.5%

In dermatoglyphics, statistically significant findings are, absence of elliptical whorl pattern in finger tip patterns of both hands and absence of tibial loop in area V of ball region of left foot in patients as compared to controls. Conclusions: Absence of elliptical whorl pattern in finger tip patterns of both hands and tibial loop in area V of ball region of left foot in dematoglyphics while microdeletions (2)(q12);(10)(q25) in chromosomal study are significant findings in PCOS which are co-related and can be used as marker. Chromosomal abnormalities might be the cause of failures in ARTs in such cases. This would be of great help to infertility clinicians for screening and to get a guideline to decide line of treatment while to the patients to reduce psychosocial and financial stress during treatment.

1. Introduction

Polycystic Ovary Syndrome (PCOS) is one of the most common cause of infertility in women of child bearing age. Polycystic ovaries are commonly detected by ultrasound or other forms of pelvic imaging, with estimates of the prevalence in the general population being in the order of 20–33%.1, 2 The genetic mechanisms underlying the PCOS remain largely unknown. Given the large number of genetic variants found in association with these disorders, the emerging picture is that of a complex multigenic trait.

Chandley3 (1998) quoted that karyotype should be performed for infertile women suspected of having chromosomal aberrations. Now a days, cytogenetic screening of both the partners is mandatory prior to any type of Assisted Reproduction Techniques (ARTs) in some countries such as Germany, UK, USA, Kuwait, Turkey and Spain. Much of the work done in India is mainly in male infertility.

Franks S4 (1999) proposed that PCOS is an oligogenic disorder in which a small number of key genes interact with environmental factors (notably dietary), the balance of which factors determine, the typically heterogeneous, clinical and biochemical phenotype.

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Dermatoglyphics (derma – skin, glyphos – carving) is the study of epidermal ridge patterns of palms, fingerprints and soles. Wide spread medical interest has been developed in dermatoglyphics in last few decades. The role of dermatoglyphics as a diagnostic tool in screening genetically transmitted diseases is becoming increasingly popular since,Ridge configuration is genetically determined. Chromosomal aberrations alter the ridge patterns.Ridges are formed in the 18th week of IUL and remain the same throughout life. Meenakshi S.5et al. (2006) studied dermatoglyphics of amenorrhea. This is an attempt made probably for the first time to study dermatoglyphics in infertile female patients with PCOS.

Aims and objectives:

1. To study the chromosomal and dermatoglyphic patterns of infertile female patients with PCOS.

2. To try and co-relate dermatoglyphics with chromosomal aberrations in PCOS.

3. To look for a marker for mass screening in female infertility.

4. To try and co-relate chromosomal aberrations and repeated failures after ARTs in patients with PCOS which will help to decide about the line of treatment.

Material and methods:

16 infertile female patients with clinically diagnosed PCOS according to the Rotterdam6 ESHRE / ASRM revised 2003 consensus on diagnostic criteria3were selected along with same number of fertile normal females as control.

Chromosomal study was pursued by collecting 2 ml of blood in heparinized syringe under aseptic conditions and incubating it with culture medium for 68 hours in BOD incubator at a fixed temperature of 370C and processed further in genetic laboratory. The chromosomes were stained by Giemsa banding technique7. Chromosomes were identified and arranged in groups A to G so also X and Y chromosomes as per their characters and the configuration of light and dark bands over them. Karyotypes were analyzed for chromosomal abnormalities.

Dermatoglyphic study was undertaken by using roller ink method with Kore's duplicating ink. Prints of both palms of hands and soles of feet were taken. Patterns were analyzed using Henry's system as

· Arches (simple(F1) and tented(F2)),

· Loops (double(F4), ulnar(F5), radial(F6), central pocket(F7)),

· Whorls (spiral(F9),concentric(F10), elliptical(F11), 'S' shaped(F12), central pocket(F13),)

· Composite(F8) and

· Vestige pattern (F3) for both palmar and plantar dermatoglyphics.

The palmar and plantar dermatoglyphic patterns so also the karyotypes were compared with that of controls to know if they are differing from the normal patterns and were statistically analyzed.

Observations:

Chromosomal study revealed,

Incidence of chromosomal aberrations in infertile females with PCOS is 81.25%.

Further distribution of different chromosomal abnormalities found is as follows:

Microdeletion Observations:	Homozygous/ Heterozygous	Incidence (%)	Photograph no
2q12	homozygous	50	1
2q12	heterozygous	25	2
10q25	heterozygous	12.5	2

(Figure 1, Photograph 1& 2)

Autosomal translocation found was der(7) t (14;7)(q21→ 31;q36) heterozygous i.e. translocation of region q21→ 31on chromosome 14 to chromosome 7 at q36 which is 06.25%. (Photograph 3)

Numerical chromosomal abnormality was observed as 47XXY female in 06.25% of cases. (Photograph 4)

Dermatoglyphic study revealed

Following significant findings were found in patients with PCOS as compared to controls: A. Palmar dermatoglyphics:

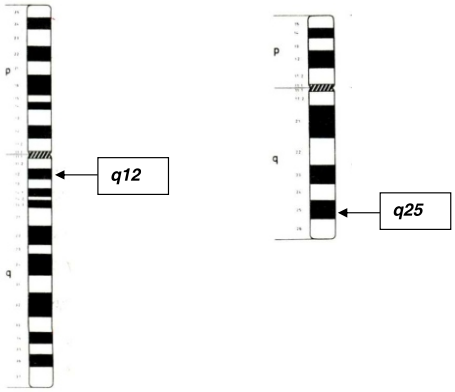
Elliptical whorl pattern in finger tip patterns of both the hands is absent in PCOS and it is statistically significant. (photograph 5)

B. Plantar dermatoglyphics:

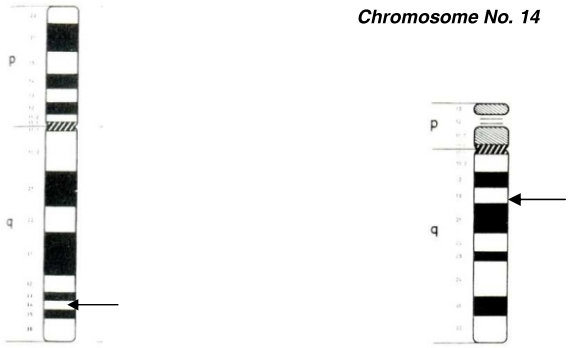
Tibial loop in area V of ball region of left foot is absent in PCOS and it is statistically significant. (photograph 6)

Figure 1[5] (Ideograms of G-Banding patterns)

Chromosome No.2 ***Chromosome No.10***

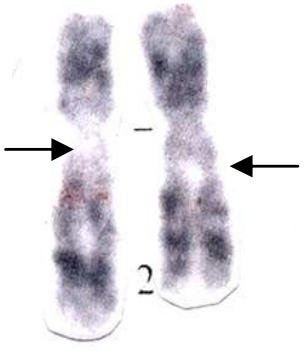


Chromosome No. 7 ***Chromosome No. 14***



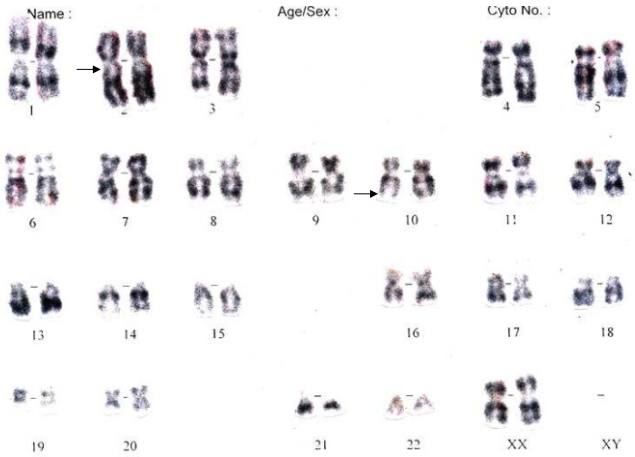
Karyotypes:

Photograph 1



Chromosome 2 showing: -, deletion (2)(q12)homozygous.

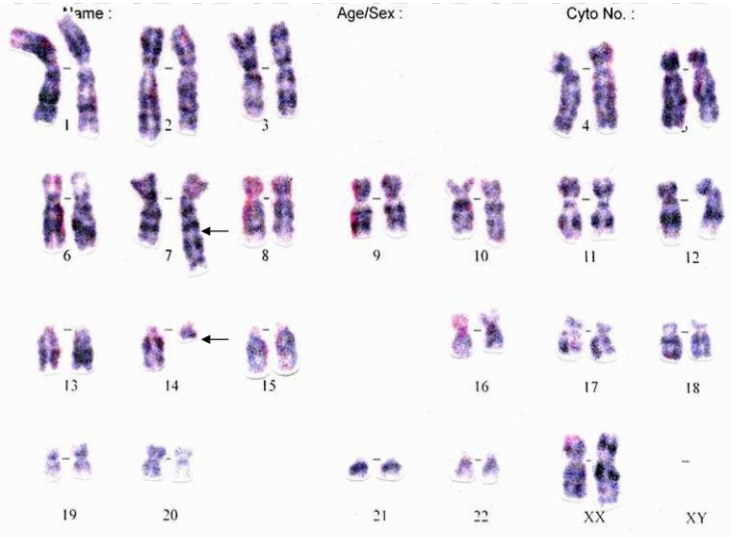
Photograph 2



Karyotype Findings:-

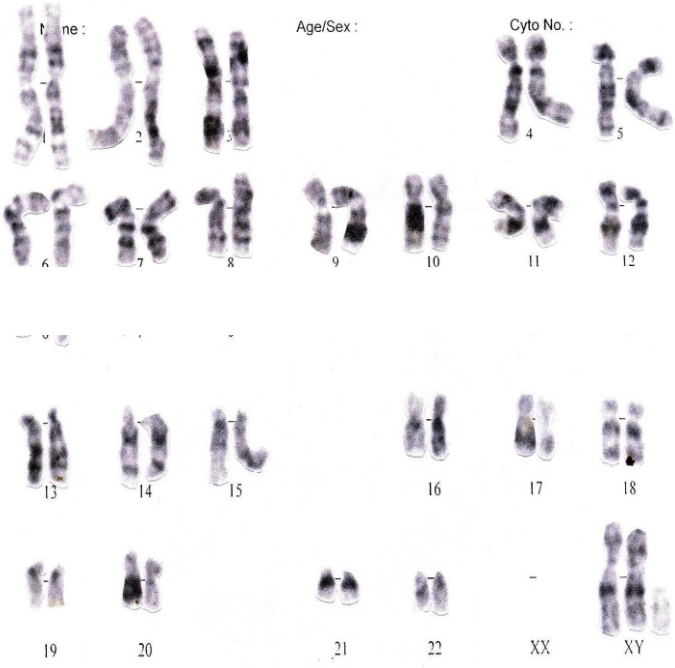
46,XX del(2)(q12) heterozygous, del(10)(q25) heterozygous.

Photograph 3



Karyotype Findings: - 14-7 Translocation
46,XY der (7) t (14;7) (q21 - 31;q36) heterozygous

Photograph 4



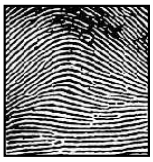
F11.Elliptical Whorl F12. 'S' shaped Whorl F13. Central Pocket Whorl

Photograph 4

Karyotype Findings: -

47,XXY.

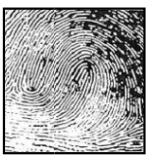
Dermatoglyphic Patterns:



F1. Simple Arch Whorl

F2.Tented Arch

F3.Vestige Pattern



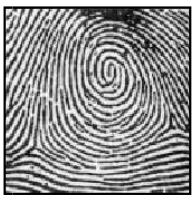
F4.Double loop

F5. Radial Loop

F6. Ulnar Loop

F7.Central pocket Loop

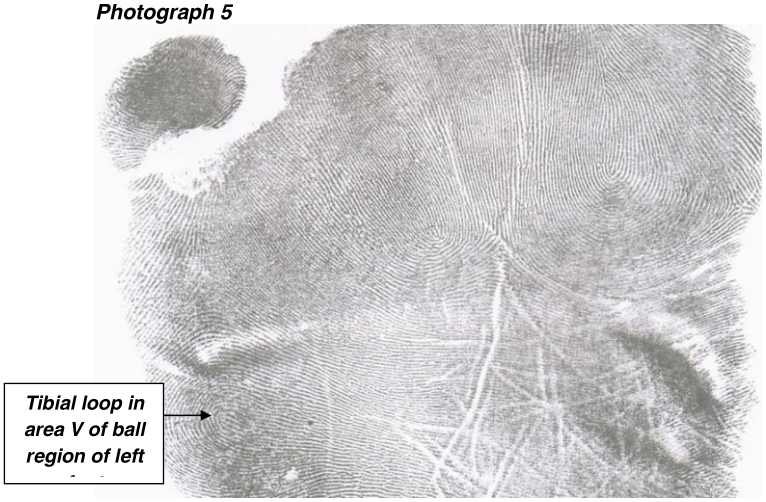
F8. Composite Patterns



F9. Spiral Whorl

F10. Concentric Whorl

Photograph 5



Discussion:

Andrea8 (2001) stated that family studies have indicated a genetic susceptibility to PCOS. Family-based studies of linkage and association have implicated several genes in the pathogenesis of PCOS and the strongest evidence to date points to a gene in the region of the insulin receptor.

Nersesyan A9 (2006) concluded that females with PCOS have increased chromosomal aberrations (CAs) level in lymphocytes which is a sign of genetic instability.

Urbanek M. 10 (2007) mentioned that during the past decade, the roles of more than 70 candidate genes have been evaluated for a causal role in PCOS; however, because of genetic and phenotypic heterogeneity and underpowered studies, the results of many of these studies remain inconclusive.

In present study 81.25% of infertile females having PCOS are showing chromosomal aberrations either structural or numerical. Few of those infertile females with PCOS which showed chromosomal abnormality, suffered from repeated failures of ARTs. May be this is the time to think whether the chromosomal abnormalities in these patients is the cause of failures of ARTs in spite of best of the treatment. If it is so it might give a guideline to the treating doctors to decide about the line of treatment and might also help the patients to avoid financial and psychosocial stress of failure in treatment.

Dermatoglyphic and chromosomal findings have definite co-relation because dermatoglyphic patterns are under genetic influence. Modern cytogenetic method allows precise identification of chromosomes and thus helps in studying the co-relation between individual chromosomal aberrations and dermatoglyphic features. Due to this, diagnostic role of dermatoglyphic patterns is bright especially in chromosomal abnormalities. Above dermatoglyphic findings might help to get a guideline to know whether female patients with primary infertility have PCOS.

Conclusion:

Present study shows that there is definite co-relation between chromosomal abnormalities, dermatoglyphics and PCOS. Elliptical whorl pattern in finger tip patterns of both the hands is absent as well as Tibial loop in area V of ball region of left foot is absent in PCOS. Since dermatoglyphics is a cost effective, less time consuming and easier method for screening which can be used anywhere without stationary lab equipments, it may be of great help in screening the population for PCOS.

Chromosomal study revealed significant percentage of chromosomal abnormalities in patients with PCOS like del 2q12 homozygous in 50% (8/16), del 2q12 heterozygous in 25%cases (4/16), and del10q25 heterozygous in 12.5 % (2/16) so also 14/7 translocation and 47XXY female are of importance. This would be of great help to come to some conclusion about the possibility of cause of infertility and failures of ARTs or repeated abortions in case of ARTs. From the result of present study, it seems that now there is a time to think whether chromosomal study should be made compulsory before proceeding for ART in infertile patients to avoid psychosocial and financial stress to the couples.

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