Original Article

Genetics Section

Pedigree Analysis and Cytogenetic Study in Vitiligo

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ABSTRACT

A total 300 cases of vitiligo with family history & cytogenetic study in patients having positive family history male to female ratio 1:1. Vitiligo begins in the younger age groups, symmetrical lesions in 50% cases, 72 cases gave definite positive family history & transmitted by autosomal dominant characteristic.

Key Words: Vitiligo, Multilocus recessivity, Segregation analysis

INTRODUCTION

Leucoderma is a general term applied to decreased melanin pigmentation of skin. Vitiligo is the commonest type of leucoderma. Daniel Turner (1714) was the first British dermatologist, who recognized vitiligo. Vitiligo is an idiopathic, hypo melanositic, dermatological disorder characterized by pale milky white macules that tend to become progressive over time. It is not present at birth but develops later in life [1, 2, 3].

The disease is characterized by loss of normal colour of the skin in patches, resulting in various degrees of cosmetic disfigurement. It may cause a severe sense of humiliation, worry, anxiety & represent a social obstacle throughout life. The affected patient prefers seclusion & abstains from appearing in public, lest he should be looked upon as having a contagious disease [4].

The oldest information concerning it comes from Pharonic Medicine in the Ebers papyrus. The Indian sacred book Atharva Veda mentions a disease known as “shweta-kustha” [5]. In Arabic language bohak, bahak, baras are Arabic names for vitiligo. The term vitiligo, derived from the Latin word ‘vitelius’, means calf. It was first used by the Roman physician Celsus.

Genetic control of pigmentation in mammalian systems is complex. At least 147 genes at 53 different loci affect coat colour in mice. Inherited pigmentary defects in humans are known to involve mutations in genes which control various steps in the pigmentary process. In general, mutations which interfere with the development & migration of melanoblasts to peripheral sites are frequently associated clinically with localized hypopigmentation & deafness [6, 7, 8].

While those mutations which interfere with melanosome formation and the synthesis of melanin result in defects with the general clinical characteristics of oculocutaneous albinism.

AIMS & OBJECTIVES

- Family study [pedigree analyses] of vitiligo patients.
- Cytogenetic study of patients having positive family history of vitiligo.
- To find out whether or not any chromosomal abnormality exists in vitiligo patients.

MATERIALS & METHODS

The present study, carried out in Department of Anatomy, GMC Nagpur, comprises of pedigree analysis [family study] & cytogenetic study of 300 cases of vitiligo patients from Skin OPD, clinically diagnosed cases of vitiligo were selected. Patients were studied with age, sex, geographic area, occupation, education status, food habits includes alcohol intake, family history up to 3 generations, history of allergy, size, shape, symmetry, distribution, pattern & colour of borders in vitiligo [9, 10, 11].

Chromosomal studies were carried out on blood lymphocyte culture. Peripheral blood for culture was drawn from the median cubital vein under complete aseptic conditions, in a heparinized syringe. Then 3-4 drops of blood was dropped into culture bottles containing RPMI 1640 medium. Culture bottles were incubated at 37°C for 72 hrs with stopper tightly closed. To arrest mitosis colchicine was added to medium 2 to 3 hrs before harvesting. The cultures were centrifuged twice at 1000 RPM for 5 minutes. Phytohaemagglutin was added to swell the cells & they were fixed in alcohol at room temp for 30 minutes. The supernatant was discarded completely without disturbing the cells at the bottom. Then 2-3 drops of cell suspension were dropped on wet, ice cold, grease free slides from a distance, to facilitate spreading. Further the slides were stained by Giemsa stain. Karyotyping of the slides was done using photokaryotyping. A camera was attached to the microscope. Photographs of the metaphase spread were taken. From the photograph of each metaphase spread, the chromosomes were cut out & aligned according to centromere position. In this manner the karyotypes were prepared for all 50 cases [7, 8].

OBSERVATION

Age	Male	Female	Total	%
10-20yrs	60	42	102	34%
21-30yrs	53	32	85	28.33%
31-40yrs	20	32	52	17.33%
41-50yrs	8	29	39	13%
51-60yrs	9	15	22	7.33%
Total	150	150	300	100%

[Table/Fig-1]: Distribution of patients by age

	Number of cases	%
Mother	22	7.33%
Father	18	6%
Sister	10	3.33%
Son/daughter	11	3.66%
Uncle/aunt	3	1%
Grand parents	5	1.66%
Cousins	3	1%
More than one relations	10	3.33%

[Table/Fig-2]: Distribution of patients relatives having vitiligo

Size of macules	No of cases	%
1cm	25	8.33%
2-5cm	200	66.66%
6-10cm	52	17.33%
>10cm	23	7.66%
Total	300	100

[Table/Fig-3]: Size of vitiligo lesions
Out of 300 cases 72 cases [24%] gave definite family history

Status	Duration in yrs			Total	%
	> 2	2–10	>10		
Stationary	50	13	10	73	24.33%
Progressive	85	130	12	227	75.66%
Total	135	143	22	300	100

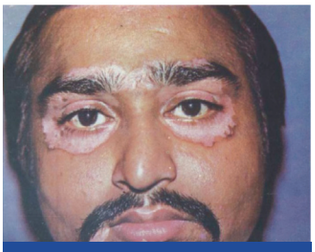
[Table/Fig-4]: Duration & status of disease

Colour of borders	No of cases	%
Hyperpigmented	115	38.33%
Erythematous	2	0.66%
Normal	183	61%
Total	300	100

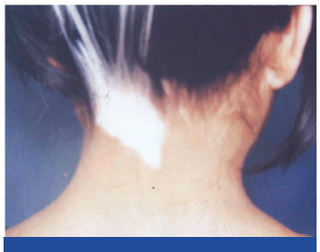
[Table/Fig-5]: Colour of vitiligo borders

	Ass. Diseases	No of cases	%
1	Ischaemic heart dise.	11	3.66%
2	Cervical spondylosis	9	3%
3	Bronchial asthma	10	3.33%
4	Tuberculosis	2	0.66%
5	Diabetis mellitus	20	6.66%
6	No systemic dise.	199	66.33%
7	Psoriasis	9	3%
8	Pemphigus vulgaris	1	0.33%
9	Lichen planus	6	2%
10	Eczema	22	7.33%
11	Alopecia areata	2	0.66%
12	Syphilis	1	0.33%
13	Not associated with skin disease	259	86.33%

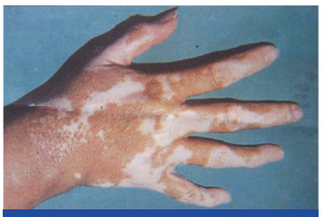
[Table/Fig-6]: Concurrent dermatological and systemic diseases in vitiligo patients



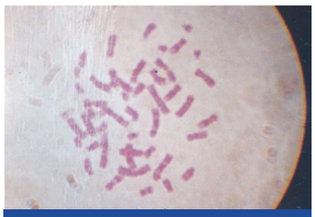
[Table/Fig-7]: Orofacial vitiligo



[Table/Fig-8]: Generalised vitiligo



[Table/Fig-9]: Hand vitiligo



[Table/Fig-10]: Metaphase spread

DISCUSSION

The following observations were found in present study.

In our study, vitiligo appears in younger age group. The male: female ratio was 1:1 [Table/Fig-1 & 6].

The commonest size of vitiligo lesions ranged from 2 to 5 cm in 200 cases, and symmetrical lesions were observed in 50% cases [Table/Fig-8 & 9].

FAMILY STUDY

72 cases [24%] out of 300 gave a positive family history [Table/Fig-2].

Out of 72 cases, 10 [3.33%] had more than one vitiligenous blood relation. In the remaining 62 cases, [20.66%] the patient had only one relation suffering from the disease. Of these, 22 [7.33%] had a vitiligenous mother, 18 [6%] had a vitiligenous father, 10 [3.33%] had a vitiligenous sister, 11 [3.66%] had a vitiligenous son/daughter, 3 [1%] had a vitiligenous uncle /aunt & 3 [1%] had vitiligenous cousins [6, 9, 10].

Several authors reported similar findings.

Status of disease: In 227 cases [75.66%] the disease was progressive & remained stationary in 73 cases [24.33%].

CHROMOSOMAL ANALYSIS

Chromosomal analysis was carried out in the 70 patients who had positive family history. Metaphase spreads could be studied in 50 cases [71.42%] since cultures failed in 20 cases [28.57%]. The karyotype was normal; there were no structural & numerical abnormalities [11, 12] [Table/Fig-10].

CONCLUSION

Vitiligo presents itself with onset at an early age. The lesions tend to be small in size yet progressive. Vitiligo is more evident in early ages of males (upto 30 years of age) than females. Vitiligo follows a polygenic, multifactorial inheritance pattern. However, none of the karyotypes presented structural or numerical abnormalities. Significant family inheritance presents itself as an appreciable factor for this disease. Chromosomal changes eg metaphase are evident in half of cases.

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