Asian Resonance

A study of Numerical aberration of chromosomes in albinism cases

Abstract

Leukocyte cultures were carried out from 15 normal individuals and 16 cases of albinism. Present research was to study changes in chromosomes of albinism cases. The frequency of numerical chromosomal aberration in albinism cases are not much increased compared to normal individuals

Keywords: Numerical Chromosomal aberration, albinism.

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Introduction

Albinism is an inherited disorder of melanin synthesis affecting skin hair and eyes (Wiktop 1978). Such a condition is found occasionally in many species of mammals. Depigmentation is present from birth. Great majority of albinos are the offspring of parents who are normal in appearance. These person are sensitive to bright light and their skin is easily sun burned. Melanin, haemoglobin and carotene are the three pigments that impart a variety of colours to skin (Goldsmith, Mckusick 1979).

Melanocytes synthesize melanin from the amino acid tyrosine in the presence of enzyme tyrosinase (Totora, Derrickson 2009). Synthesis occurs in an organelle called melanosome. Exposure to ultra violet light increases the enzymatic activity and melanin production within melanosomes. Amount and darkness of melanin increase upon ultra violet exposure which gives a skin tanned appearance. Melanin absorbs ultraviolet radiation and prevents damage to deoxyribonucleic acid in epidermal cells and neutralizes free radicals that form in the skin following damage by ultraviolet radiation. But exposing the skin to a small amount of ultra violet light is necessary for vitamin D synthesis. Albinism is the inherited inability of an individual to produce melanin. Skin and hair colour are controlled by numerous genetic loci in humans. Lack of pigment in the skin makes albinos sensitive to sunlight increasing the incidence of skin, burns, lack of pigment in the eyes may contribute to photophobia (Agrawal et. al, 2007).

Amongst all types of metabolic genetic abnormalities albinism is less lethal. In majority of people it is inherited in the form of recessive character. This is due to abnormality in the metabolism of phenyl alanine tyrosine caused on account of abnormal recessive metabolism. Disease is caused by recessive gene called as albino gene. It appears when it is in homozygous condition. There is lack of tyrosinase in enzyme system which is required for the cinversion of 3, 4 dihydroxy phenyl alanine into melanine pigment inside the melanocytes. Melanocytes are present in normal number in albino patients. In albinism even though the metabolic block leading to an abnormality is known, it is difficult to correct it (Devtin 2005). (Rencuzogullari, Kayraldiz, 2001) studied chromosome aberrations and sister chromatid exchange in cultured human lymphocytes treated with sodium metabisulphate.

Material And Methods

Peripheral leucocytes cultures were initiated to obtain a large number of cells in metaphase stage. Dalhousie University, Halifax Novaseatia method requires only few drops of blood. 8 to 10 drops of blood were dropped directly into each of universal containers having following : 5 ml of Tc 199 medium, 1 ml of serum, .15 ml PHA (Phytohaemaglutinin) and 1 – 2 drops of heparin. Culture bottles were kept in water bath at 37^{O} C for 72 hrs 0.3 ml Colchicine (0.04%) was added to each culture tube on morning of third day. After giving hypotonic treatment, cells were fixed in freshly prepared fixative. Giemsa was used for staining slides. All the stained slides from each aliquot were labelled and screened under lower power for the quality of preparation. Slides were selected for differential count of the metaphases. The chromosome analysis were made on

microscope with film projection at magnification. Twenty metaphases were analyzed for each case.

Results And Discussion

Leucocyte cultures were initiated from 15 normal individual. Age of these 15 individual with positive cultures ranged from 21 to 45 years. They were 9 males and 6 females.

Table - 1
Showing sex – wise distribution of 15 healthy individuals

S. No.	Sex	No. of cases
1.	Male	9
2.	Females	6
	Total	15

Table – 2
Showing distribution of cases according to age

S. No.	Agewise distribution	No. of cases
1.	21 to 25 years	5
2.	26 to 30 years	4
3.	31 to 35 years	4
4.	36 years onwards	2
	Total	15

Table 3 shows numerical aberrations in only eight cases hypoloid metaphases were observed. .

Table – 3
Showing numerical aberrations in normal individuals.

S. No	Cases number	Hyperploid metaphase	Hypoploid metaphase	Chromosomal pattern of abnormal metaphases
1.	N-1	-	-	-
2.	N-5	-	-	-
3.	N-5	-	-	-
4.	N-6	-	-	-
5.	N-7	-	-	-
6.	N-10	-	3	45 XY – C
7.	N-12	-	-	-
8.	N-15	-	-	-
9.	N-16	-	5	45 XX – B
10.	N-18	-	-	-
11.	N-19	-	-	-
12.	N-20	-	-	-
13.	N-22	-	-	-
14.	N-23	-	-	-
15.	N-28	-	-	-

Chromosome groups affected by aneuploid metaphase in normal individuals

Table - 4 shows that chromomes of group – B and Group C were affected in normal individuals, whereas chromosomes of other groups were not affected. Chromosomes of group – B were more affected.

Table – 4 :
Showing chromosomes group affected by aneuploid metaphase

Chromosomes groups	Hyperploid	Hypoploid
A	-	- -
B	-	5 -
C	-	3 -
D	-	- -
E	-	- -
F	-	- -
G	-	- -

Distribution of metaphases with numerical aberrations according to sex in normal individuals

Male – One hundred and eighty metaphases were analyzed from 9 normal males. Three metaphases exhibited numerical aberrations in 01 cases represented by hypoploid.

Female – One hundred and twenty metaphases were analysised from 6 females. Five metaphases exhibited hypoploidy in one case.

Table – 5
shows sexwise distribution of aberrations in normal individuals

Sex	No. of cases	Total No. of meta-phases analyzed	No. of cases showing abnormal meta-phases	Hyper-diploid meta-phases	Hypo-diploid meta-phases
Male	9	180	2	-	3
Female	6	120	1	-	5

Age-wise distribution of numerical aberrations in controls

In the age group 20 to 25 years. 100 metaphases were analyzed. Only 8 metaphases exhibited hypodiploiy in 2 cases. In the age group 25 to 30 years, 80 metaphases were analyzed, in the age group 30 to 35 years, 80 metaphases were analyzed. No numerical aberrations were found. In the age group 35 to 40 years. 40 metaphases were analyzed. No numerical aberrations observed. Results are summarized in Table – 6.

Table– 6
Showing distribution of metaphases with numerical aberrations according to age in normal individuals.

Age in years	No. of cases	No. of meta-phases analyzed	Number of cases showing abnormal meta-phases	Numerical aberrations			
				Hyper-diploidy		Hypo-diploidy	
				No.	%	No.	%
20 – 25	5	100	2	-	-	8	8.0
26 to 30	4	80	-	-	-	-	-
31 to 35	4	80	-	-	-	-	-
36 onwards	2	40	-	-	-	-	-

Albinism Cases – Leucocyte cultures were performed from 16 cases of albinism. Positive culture results were obtained in eight cases. They were 4 male and 4 female. Age of these cases varied from 8 to 60 years.

Table – 7
Shows distribution of cases according to sex

S. No.	Sex	No. of cases
1.	Male	4
2.	Female	4
	Total	8

Table – 8
Shows distribution of cases according to sex

S. No.	Age groups in years	No. of cases
1.	8 to 20	2
2.	21 to 32	2
3.	33 to 44	2
4.	45 to 56	1
5.	57 to 60	1
	Total	8

Numerical aberrations

In case number A-3, two metaphase showed hypoploidy. In case number A-6, one (4%) metaphase showed hyperploidy and 5 (20%) metaphase showed hypoploidy (Table No. 9).

Table - 9
Shows numerical aberrations in a study on 8 cases of albinism.

S. No	Cases number	Numerical aberrations			Chromosomal of abnormal metaphases
		Hyperploidy	Hypoploidy	Other types	
1.	A-1	-	-	-	-
2.	A-2	-	-	-	-
3.	A-3	-	2	-	45, XX-D
4.	A-4	-	-	-	-
5.	A-5	-	-	-	-
6.	A-6	1	5	-	Five 45, XX-B One 47, XX+E
7.	A-7	-	-	-	-
8.	A-8	-	-	-	-

Table – 10
shows distribution of numerical aberrations according to affected chromosomes groups

S. No.	Chromosomes group	Hyperploidy	Hypoploidy
1.	A	-	-
2.	B	-	5
3.	C	-	-
4.	D	-	2
5.	E	1	-
6.	F	-	-
7	G	-	-

Distribution of numerical aberrations according to age

In age group 8 to 20 years, fifty metaphases were analyzed pattern, in age group 21 to 32 years; fifty metaphases were analyzed. None of these metaphases exhibited aneuploidy. Age group 33 to 44 years, fifty metaphases were analyzed. One metaphase exhibited hyperdiploidy and seven metaphases exhibited hypodiploidy. In age group 45 to 56 years; twenty five metaphases were analyzed, in age group 57 onwards; twenty five metaphases were analyzed. None of the metaphases exhibited numerical aberrations.

Results are tabulated in Table - 13.

Table - 11
Shows distribution of numerical aberrations according to age.

S. No	Age group in year	Number of metaphases analyzed	Numerical aberrations			Total	
			Hyper-diploidy	Hypo-diploidy	Other types	No.	%
1.	8 to 20	50	-	-	-	-	-
2.	20 to 32	50	-	-	-	-	-
3.	33 to 44	50	1	7	-	8	16.0
4.	45 to 56	25	-	-	-	-	-
5.	57 onwards	25	-	-	-	-	-

Distribution of numerical aberrations according to sex

Female – one hundred metaphases were analyzed in 4 females. Eight metaphases showed aneuploidy in two cases.

Male – 100 metaphases were analyzed from four males. All showed normal chromosomal pattern.

Table - 12
Shows distribution of numerical aberrations in 8 cases of albinism according to sex.

Sex. `	Number of cases	Number of metaphases analyzed	Number of abnormal metaphases	Numerical aberrations Total			
				Hyper-diploid	Hypo-diploid	No.	%
Male	4	100	-	-	-	-	-
Female	4	100	08	01	7	8	8

P $\angle$ 0.01 chromosomal aberrations are significance in females in female.

Albinisms is due to abnormality in the metabolism of phenylalanine tyrosine and is caused on account of abnormal recessive metabolism. Various metabolic methods are regulated by gene action. There is no primary treatment available for albinism. Genetic counseling is an essential part of the management of patient.

Leucocyte cultures were initiated from 15 normal individuals. They were 9 males and 6 females Numerical aberrations were observed in only 2 cases. 8 metaphases showed numerical aberrations (Beiguelman and Pisan's 1976) in there study analyzed 400 metaphases in the control group and found that 4 (1%) showed numerical aberrations. Thus, in the present study the numerical aberration were higher than reported by (Beiguelman and Pisani 1976). Percentage of aneuplides in the present study was lesser than that reported by Bloom et. al 1973. In the present study meteaphases showed hypoploidy no hyerploidy was observed. Chromosomes affected belonged to group B and C.

Leucocyte culture were done from blood of 16 cases of albinism. Eight cases gave positive cultures. There were 4 males and 4 females. In the

study of Cervenka et. al.(1979) 14 Cases were taken for chromosomal analysis. There were 9 males and 5 females. Age of the patients in the present study ranged from 2 to 60 years. The chromosomal analysis performed of healthy and albinism cases showed that frequency of metaphases with chromosomal aberrations are not increased in albinism cases.

The Chromosomal counts performed on the leucocyte metaphases of the healthy individuals and albinism cases showed that the frequency of metaphases with numerical aberrations are similar in two groups of individuals. Statistical analysis of the result shows that there was no significant increase in aneuploidy in albinism cases. Eight albinism cases were taken for the study of chromosomal counts. Metaphases showing abnormal counts were observed only in 2 cases. Six cases showed normal chromosomal counts. Affected chromosomes belonged to group D, B and E whereas in normal individuals chromosomes affected were of group B and C.

All males and 50% females of albinism showed normal chromosomal counts. Thus the frequency of aneuploidy in anbinism patients are much lower then the average frequency of aneuploidies found in leucocyte cultured of healthy individuals (Bloom 1972). It can be concluded that in cases of albinism chromosomal aberration are not significantly increased. (Bloom et al.1973). Aberrations found in females requires further and larger series study. Cervenka et. al, (1979) analyzed 1404 metaphase from 14 cases and found 2.13% metaphases showing chromosome aberrations. In their study the frequency of metaphases showing chromosome aberrations was lower than that in normal individuals. Increased chromosomal breaks in lymphocytes occurred in Puerto-Riean albinos with Hermansky Pudlak syndrome. (Maurer et. al, 1968,) but this was not found in albinos residing in the Northern climate of Minnerota (Wiktop et. el., 1973).

Acknowledgement

We are thankful to professor and Head Medicine Department and Dean M.G.M. Medical college, Indore for providing laboratory facilities during tenure of this research work.

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