

**INCIDENCE OF DOWN'S SYNDROME WITH CHROMOSOMAL PATTERN IN THE EASTERN INDIA**

Jami Sagar Prusti<sup>1</sup>, Sitansu K. Panda<sup>2</sup>, Santosh K. Sahu<sup>3</sup>, Dharma N. Mishra<sup>4</sup>, Geetanjali Arora<sup>5</sup>, Mahesh C. Sahu<sup>6</sup>

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**ABSTRACT: BACK GROUND:** To study the chromosomal pattern of the Down's syndrome patients of the eastern Indian population coming to the MKCG Medical College, Berhampur, Odisha, India. Methodology: 0.8ml of peripheral blood was collected from all the patients and cultured in RPMI 1640 medium for 72h in CO<sub>2</sub> incubator. Then cells were harvested with colchicines and after the hypotonic treatment the slides were prepared and stained with giemsa. After that it was observed with high power microscope and reported. **RESULT:** From the 50 patients, 39 patients were Primary trisomy 21 (47, XX or XY+21), 6 was Mosaic Trisomy 21 (46/47, XX or XY+21), 3 was Down's syndrome Primary amenorrhoea (47, XX, +21) and there was 2 translocation 46, XY, t (14:21) 46, XY, t (21:21). The paternal age at 26-30 years were found more whereas the maternal age at 21-25 years was found more and males were more affected as compare to the females. **CONCLUSION:** It is tempting to speculate that, the difference in clinical features, growth retardation, abnormal dermatoglyphic patterns etc, are related to the genetic constitution of the Down's syndrome individuals.

**KEYWORDS:** Karyotype, Down's syndrome, colchicines, CO<sub>2</sub> incubator.

**INTRODUCTION:** Down's syndrome is a genetic condition that causes delays in physical and intellectual development. It occurs in approximately one in every 800 live births. Individuals with Down's syndrome have 47 chromosomes instead of the usual 46. It is the most frequently occurring chromosomal disorder.

Down's syndrome is not related to race, nationality, religion or socioeconomic status. The most important fact to know about individuals with Down's syndrome is that they are more like others than they are different.

Down's syndrome is the most common chromosomal disorder in humans. Its prevalence in Europe is about 9.8:10 000 live born infants,<sup>(1)</sup> while in the USA it is 8.5:10 000 newborns to mothers younger than 35 years of age and up to 55.3:10 000 newborns among mothers older than 35 years of age.<sup>(2)</sup> Genetic cause for this syndrome is trisomy of chromosome 21 or the presence of distal part of the long arm of chromosome 21.

In Down's syndrome patients there are three types of cytogenetic trisomy 21, i.e. free trisomy 21, mosaic trisomy 21, and translocation trisomy 21 are present.<sup>(3)</sup> Most commonly, Down syndrome children have karyotype of free Trisomy 21, while their parents have normal karyotype. This type of trisomy 21 exclusively occurs sporadically de novo as a result of non-dysjunction of homologous chromosomes 21 during gametogenesis to parents or during early embryonic development after fertilization.<sup>(4)</sup>

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Analysis of chromosome heteromorphisms and many other informative markers of DNA polymorphisms of parents and their offspring with Down syndrome revealed that chromosome 21 non-dysjunction occur more often during the gamete-formation process in females than in males.<sup>(5,6)</sup>

Investigations found that an extra chromosome 21 mainly originates from errors in maternal side in approximately 90% of the Down syndrome cases. In 5-10% of Down syndrome cases the extra chromosome 21 originates due to errors in father side, whereas in less than 5% of cases it results from nondysjunction of chromosomes during a post-zygotic mitosis in early embryonic development.<sup>(7,8)</sup> Important role for chromosome 21 non-dysjunction has also maternal age. Older woman is more likely to have a chromosome 21 non-dysjunction during oogenesis than young women.<sup>(9)</sup>

Therefore older women have a higher risk of having a baby with Down syndrome. In children with Down syndrome due to translocation trisomy 21, extra chromosome 21 is joined or translocated in any other acrocentric or non-acrocentric chromosome.<sup>(10)</sup> The translocation trisomy 21 present in Down syndrome patients can be created spontaneously de novo during gametogenesis in one of the parents or it can be inherited from parents' carrier of Robertsonian translocation or of reciprocal translocations.

Studies indicated that in the case of children having Trisomy 21 with Robertsonian translocation created sporadically de novo, the risk for a second future offspring trisomy 21 for their parents with normal karyotype is small. There is a significant increased risk of giving birth to a child with Trisomy 21 when one parent is a Robertsonian translocation carrier or of reciprocal translocations as they may produce balanced and unbalanced gametes during gametogenesis.<sup>(11,12)</sup>

When one parent is carrier of Robertsonian translocation 21q; 21q, it has 100% chance of having a Down syndrome child as all of its produced gametes are unbalanced.<sup>(13)</sup> The frequency to have one child with Down's syndrome due to translocation trisomy 21 is not influenced by the age of the mother. Peoples with mosaic Down syndrome have two distinct cell lines with different karyotype. In some cells there are a total of 46 chromosomes having normal karyotype, while other cell lines have karyotype with Trisomy in chromosome 21.<sup>(14)</sup>

Many authors argued that restricting or reducing the births for woman's who are aged 35 years or older, mandatory prenatal cytogenetics diagnosis of fetal disorders of pregnant women aged 35 years or older, and applying the methods for prenatal screening on all ages pregnant women's reduce the number of births of children with Down syndrome.<sup>(15,16)</sup>

We realized our study on the rate of birth of children with Down syndrome in eastern Indian population for the time period 2008-2010, when Kosova have had no conditions for full application of methods as described above in order to prevent birth of children with Down syndrome in our population.

**MATERIAL AND METHODS:** The material for the present study comprises of children of both sex, under the age of 20 years affected with Down's syndrome. Their intelligent quiescent from mild to severe categories drawn from school records, clinics, outdoors, indoors and institutions for mentally retarded children; of different areas covering Ganjam, Gajapati, Phulbani, Cuttack districts of Orissa and neighboring Srikakulum district of Andhra Pradesh.

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To serve as controls, age and sex matched healthy children drawn from schools were chosen for the study. For cytogenetic study, 50 (fifty) clinically diagnosed Down's syndrome cases were selected. The data were collected by using a detailed questionnaire and by personal interview of parents of Down's syndrome children. For each case, a pedigree is drawn up. Family studies were carried to find out.

5-6 ml of venous blood are withdrawn in a disposable syringe, with hundred units of Heparin already into the sterile culture bottles containing culture medium- a mixture of 7 ml of RPMI, 2 ml of fetal calf serum and 0.2 ml of PHA-M (Phytohaemagglutinin). The PH of the culture is maintained between 7.2 to 7.4. The culture is then incubated for 72 hours at 37<sup>o</sup> C in CO<sub>2</sub> incubator (Moorhead et al1960). After that 0.2 ml is added to the culture, two hours prior to harvest i.e. at 70<sup>th</sup> hour.

Culture bottle are removed from the incubator at 72<sup>nd</sup> hour and the cell suspension is transferred into a 10 ml centrifuge tubes and labeled with marking pencil. Centrifuged for 8mins at 800 rpm and supernatant is discarded with a micro pipette. Resuspend the cells in hypotonic solution (KCl of 0.075 M- pre-warmed at 37<sup>o</sup> C) and incubated for 20mins at 37<sup>o</sup> C. Centrifuged for 8 mins at 800 rpm and supernatant fluid is removed.

Resuspend the cells in 0.5-1.0 ml of resting fluid. Chilled fixative (3 parts of methyl alcohol, 1 part of glacial acetic acid) is added slowly almost drop-wise, immediately mixed gently with a pipette and kept for 10mins at the room temperature. Centrifuged for 8mins at 800 rpm and supernatant fluid is removed. Again resuspend the cells in fresh fixative and keep in the refrigerator for 2 hours. Centrifuge for 8mins at 800 rpm and supernatant fluid is removed.

Re-suspend the cells in few drops of fresh fixative; confirm the cell pellet, (clear white part). If not clear, treat with fresh fixative and repeat step 9. The supernatant fixative is removed and finally 0.5 ml to 0.75 ml fresh fixative is added to the cell button (depending on the cell density) to obtain a fairly dense cell suspension.

**Slide Preparation:** After fixation, for preparation of slides, the air-drying method (first described by Rothfels and Siminovitch, 1958) is followed. Clean slides are taken and made grease free by dipping in ethyl ether overnight. A drop of fixed cell suspension is then placed on clean slide allowing it to spread out, and then dry it rapidly. The air-drying method at most times yield a higher number of better metaphase. Once the slides are dry, they were coded properly using a pencil. The dried and coded slides were kept for 5 to 7 days inside an incubator at 37<sup>o</sup> C for maturation.

The matured slides were treated with Trypsin solution for 20 seconds, which was made in one cuplin jar (trypsin 50 mg /100 ml of distilled water). The trypsin digested slides were rinsed with (NaCl 0.9%) twice which were kept in two cuplin jars. The slides were again washed with distilled water and stained with 2 ml of Giemsa stain (4%) which was poured over the slides and kept for 1 minute after which equal amount of distilled water was added for 5 minutes.

The stained slides were washed with distilled water thoroughly and allowed for air-drying. The stained and air dried slides were examined under light microscope for screening, which shows alternate light and dark bands on the chromosomes. These slides were screened for 20 well spread metaphases, five of them were photographed and one was karyotyped and studied for chromosomal aberrations. (Standard protocol ISCN,1995). The enlarged microphotographs were taken and karyotyped manually by considering the length of the chromosome in decreasing order, position of the centromere, presence of satellite bodies and banding pattern.

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**RESULT:** The various types of digital patterns of Down's syndrome cases showed a marked difference from that of normal. It is observed that the frequency of occurrence of whorls and loops in Down's syndrome cases are 17% and 76% respectively (Table 1). The 13% decrease in whorls and 17% increase in loops in comparison to normal. In Down's syndrome, the Furuhat's index is less than half of that of normal, due to high frequency of loops in these cases. In Down's syndrome the Dank meijer's Index shows a marked increase value in comparison to normal.

The "atd" angle is generally  $40^{\circ}$  –  $50^{\circ}$ . 80% of normal cases come under the range,  $30^{\circ}$ - $40^{\circ}$ . In Down's syndrome, 50% cases come under the range,  $51^{\circ}$ -  $60^{\circ}$ . In Ring and little fingers of right and left hand of normal, there is significantly more ridge count, also in thumb, a little more, in comparison to Down's syndrome Cases. Whereas in Down's Syndrome cases, in index and middle finger there is less rise of ridge count in comparison to normal (Table 2).

No significant difference was observed in sex ratio between the Down's syndrome cases, where 34 children were males and 16 were females with sex ratio of 2.03. Among the normal sibs, the sex ratio was 2.-06. No significant difference was observed in sex ratio between these groups because of awareness of the health, literacy implementation of family planning and counseling etc. in study group (Table 3).

In this present study the mean maternal age of DS cases was estimated as 26.08yrs and that of normal children as 23.51yrs. Issac, 1985, reported maternal age of DS children as  $29.54 + 6.4$ yrs and that of normal children as  $25.75 + 2.12$  yrs (Table 4). The reason for maternal age effect is not known. Several hypotheses have been proposed in particular that of aging or over ripeness of ovum, due to delayed fertilization, maternal endocrine disturbances such as dysthyroidism (Myers, 1938) and Murphy (1947).

Data on the distribution of paternal age in study and control group are presented as 48% of DS were born to fathers above 30yrs of age.

The mean paternal age of DS cases is estimated as 30.79 yrs and that of normal is 27.21 yrs. Earlier investigators (Penrose, 1933, 1962, Siglar et al 1965, Lilenfeld and Benesch, 1969) did not observe any association of DS with increase paternal age, but Stene et al, 1977, Matsunga, 1978 and Erickson, 1978 have reported increase incidence of Down's syndrome case with advancing paternal age particularly after the age of 55 years. Recent studies show that, there is no significant association of Down's syndrome with paternal age of less than 40 years. In the present study, 2% cases are observed above 40 years of paternal age in Down's syndrome subjects (Table 5).

Karyotype abnormalities reported by various authors in Down's syndrome subjects in comparison with present study is displayed in Table - 6.

**DISCUSSION:** The present study shows the occurrence of Primary trisomy 21 (48/50, 96 %), Mosaic trisomy (1/50, 2 %), Translocation (0/50; 0%) and Dow's Syndrome with Primary amenorrhoea (1/50, 2 %) respectively. A young woman who has not attained menarche even after the chronological age of 19 years shall be regarded as having primary amenorrhoea. The phenotype includes the features like flat occiput, broad nasal bridge, long and loop ears and triangle faces. The results were compared with the data pooled from various studies and presented in Table-7. From the pooled data, it is observed that 93.10% of the Down's syndrome children had pure trisomy 21, where- as 3.00 % were mosaic trisomies and 3.88 % translocation trisomies.

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It is clear from the table 4, that the frequency of pure trisomy 21 varied 100 % (ICMR study 1984) to 83.60 % (Ambani et al, 1984). The frequency of mosaic trisomy 21 varied from a minimum of 0.67 % (Murthy et al, 1989) to a maximum of 15 % (Kolf et al, 1964). Similarly, the frequency of translocation trisomy 21 also varied from a minimum of 1.4 % (Benirschke, et al, 1963 and Papp et al, 1977) to a maximum of 8.85 % (Krishnamurthy et al, 1981, Benirschke et al, (1963), Makino (1963), Sergorich et al (1964), Verma (1978), Young et al (1980).

Similarly Kolf et al (1964), Edgren et al (1966) ICMR study (1987) could not report translocation type of Down's syndrome. On the whole, from table 4, it is presumed that the Down's syndrome phenotype results as a consequence of three types of chromosome abnormalities i.e. pure trisomy 21, mosaic trisomy, and translocation type but the frequency of these chromosome abnormalities are almost similar irrespective of the ethnic or racial background and may aid to pinpoint the environmental effects on the etiology of the Down's syndrome.

Bernhein et al (1979), Verma and Hug (1987) and Sayee and Thomas (1988) have reported that males were more affected than females. Sex ratio in DS depends upon a number of factors, like increase in maternal age, time of fertilization, number of pregnancies, abortions, live births, injuries, radiation, use of contraceptives etc. Firstly, the male fetus and neonates are more susceptible to various kinds of trauma for reasons unknown.

Secondly, the boys are more likely to be diagnosed than girls, because of aggressive nature of boys. Thirdly, in social dimensions female feticide, least care and indifferent attitude towards the malformed female babies etc play a crucial role in sex ratio of Down's Syndrome subjects and normal.

Review of literature suggests that the incidence of DS is parental age dependent and the maternal age had been incremented more often than paternal age, with the etiology of DS. Hook (1981) demonstrated that the increase in trisomy 21 is moderate among young maternal age.

In the present study, 90% of DS children were born to mothers, less than 30yrs or is Equal to 30yrs of age.

Cytogenetics studies performed on 50 Down syndrome cases revealed that translocation trisomy 21 was found in 2 cases (4%) (Table 6). Our results are roughly similar to the results of other authors. (3, 15) Most children with translocation Down syndrome are born to mothers under the age of 21-25 year; therefore not depend on maternal age. Chromosome 21 most frequently creates translocations with acrocentric chromosomes than to other non-acrocentric autosomal or sex chromosomes.

Our findings conclusively argue that as well, where Robertsonian translocation between chromosome 21 and another acrocentric chromosome have been found in 16 children, while in only one child there was present reciprocal translocation between chromosomes 8 and 21. The findings of some other authors (10) reported high presence of Robertsonian translocation 14q; 21q in children with translocation trisomy 21 (62.34%). In our study the most frequent type of translocation were the Robertsonian translocation 21q; 21q (58.8%) (Table 4).

The second most frequent (23.5%) translocation type was the Robertsonian translocation 14q; 21q. Other Robertsonian translocation types were less present in our study cases. There has been reported that in 75% of all translocation cases it may occur de novo, while in 25% of cases, it can be inherited from one carrier parent, but more frequently by the mother side (11). In our present study spontaneously de novo translocation occurred in 82.4% of children, while in 17.6% of children inherited from a carrier parent, mothers respectively. To prevent the birth of children with Down



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syndrome in translocation affected families the early detection of parent's carriers with Robertsonian translocation involving chromosome 21 is of the great importance.

In the families who have children with translocation trisomy 21 arisen de novo i.e. when the parents have normal karyotype, the risk of giving birth to a child with translocation trisomy 21 is small (1-2%). Therefore, parents of 14 children with translocation trisomy 21 occurred as de novo event studied in our paper do not have high risk of giving birth to a second child with trisomy 21.

Since an affected child with reciprocal translocation 8;21 and an another child with Robertsonian translocation 14q; 21q, have inherited translocation from their mother side, the recurrence risk is significantly much higher (10-15 %). By applying the prenatal cytogenetic diagnosis of embryos of each of these couple pregnancies can be prevented the spread of Down syndrome within a generation.

To our knowledge, all published papers for couples which are carriers of silent Robertsonian translocation involving homologous chromosomes 21q; 21q have reported 100% risk of having a child with Down syndrome and unable to have healthy baby (13). In our study a Down syndrome child inherited Robertsonian translocation 21q; 21q from a carrier mother. This family is unable to have healthy child by embryo selection through prenatal cytogenetic diagnosis due to the fact that carrier mother can only make unbalanced gametes, thus any of her child will have Down's syndrome.

The cytogenetical study conducted on 305 Down syndrome children among Albanian population of Kosovo revealed that free trisomy 21 is significantly more frequent (93.4%) than the other types of trisomy 21. Other authors also indicated high occurrence of free trisomy 21 (92-95%) in children that have Down syndrome.<sup>(3,15)</sup> It shows that this chromosomal aberration is presented equally as frequent as in rest of the world.

Since the free trisomy 21 is the most common genetic cause of birth of children with Down syndrome it is of great importance to undertake preventive measures in order to reduce the disorder incidence among human population. For prevention purposes, the etiological factors as a cause of birth of children with Down syndrome, should be known and reduction measures should be taken to minimize impact.

Studies of many authors have shown that one of the factors which increase the incidence of births with free trisomy 21 is advanced maternal ages.<sup>(15)</sup> Very often Down's syndrome babies are born to mothers who are over 35 years of age. The results of our paper have also confirmed that a woman over 35 years of age to have highest incidence of giving birth to a child with Down syndrome (Figure 2). A relatively simple way to reduce birth incidence of Down's syndrome is the limitation or reduction of the number of pregnant women older than 35 years.

The frequency of birth of children with Down syndrome is expected to be reduced up to 20-45% with the birth limiting to mothers aged over 35 years.<sup>(15)</sup> Prenatal preventive diagnostic tests for Down syndrome in modern medicine can be realized through the application of either non-invasive tests (ultrasound and biochemical screening) or by invasive diagnosis methods such as chorionic villus sampling, amniocentesis, cordocentesis.

Antenatal careening using different biochemical markers and ultrasound are made routine in developed countries, where their national prenatal care policies obliges amniocentesis to all pregnant women aged 35 years or over, as well as maternal serum screening for younger women. To prevent the birth of child with Down syndrome recently there are considered some other preventive strategies, such as: pre-implantation genetic diagnosis (PGD) and folic acid supplementation.<sup>(15)</sup>

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As in Kosova currently there are no conditions for application of the above mentioned methods to prevent the birth of child with Down syndrome, during our cytogenetics study in children there was not observed a gradual decrease in the frequency of birth of Down syndrome among Albanian population of Kosova in 2000-2010 (Figure 1).

**CONCLUSION:** The present study reveals that the occurrence of Simian crease is about 40% in Down's syndrome cases. The increase in the frequency of ulnar loops, reduction in the frequency of whorls reduced total ridge count (TRC) and indices like Furuhtes index (22.89) and Pattern intensity index (11.0), has decreased values when compared to the normal. Though the loop percentage is very high, the Furuhtes index is less than half of normal.

It is thought that, the decreased 13% of Whorls is compensated by an increased 17% of Loops. The mean ridge counts of individual fingers and total mean ridge count of Down's syndrome cases are less than that of normal. In Down's syndrome cases, due to the distal placement of axial ('t') triradius, the 'atd' angle is significantly more.

The genetic forms of Down's syndrome are trisomy 21, Mosaic trisomy 21 and translocation 21. Most individuals (96%) with trisomy 21 have 3 copies of chromosome 21. Over 95% of the cases are caused by non-disjunction arising from either at first or second meiotic division's maternal or paternal origin. The present study shows the occurrence of trisomy 21 (48/50, 2%), translocation trisomy (0/50, 0%) and Down's syndrome with primary amenorrhea (1/50, 2%).

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Pattern type	Study group		Control group	
	number	percentage (%)	number	percentage (%)
WHORL (W)	17	17	30	30
LOOP (L)	76	76	59	59
ARCH (A)	7	7	11	11
Furahates Index (W/LX100)	22.89		50.84	
Pattern Intensity Index (2Xw + L)/n	2.2		2.38	
Dank meijer's Index (A/WX100)	41.17		36.66	

Table 1: Frequency of digital patterns and their indices in down's syndrome cases (n = 50) and controls (n = 50)

RANGE	STUDY GROUP		CONTROL GROUP	
	No. of Cases	Percentage	No. of Cases	Percentage (%)
30° - 40°	5	10	40	80
41° - 50°	15	30	5	10
51° - 60°	25	50	5	10
61° - 70°	5	10	0	0

Table 2: Range distribution of "atd" angle in down's syndrome cases (n = 50) and control group (n = 50)

Sl.no	Subjects	Male	Female	Total	Sex ratio (m/f)
1.	DS	34	16	50	2.12
2.	Normal children in DS family	23	11	34	2.09
3.	Control group	35	25	60	1.40

Table 3: Sex ratio in down's syndrome subjects (n = 50), normal children in down's syndrome families and control

DS = DOWN'S SYNDROME



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Sl. no	Age group	Study group		Control group	
		number	percentage	number	percentage
1.	15-20	4	8	3	6
2.	21-25	23	46	36	72
3.	26-30	18	36	9	18
4.	31-35	3	6	2	4
5.	36-40	2	4	0	0
6.	>40	0	0	0	0

Table 4: The distribution of maternal age in study group (n = 50) and control group (n = 50)

Sl. no	Age group	Study group		Control group	
		Number	Percentage	Number	Percentage
1.	21-25	3	6	4	8
2.	26-30	23	46	41	82
3.	31-35	18	36	5	10
4.	36-40	5	10	0	0
5.	>40	1	2	0	0

Table 5: The distribution of paternal age in study group (n = 50) and control group (n = 50)

Sl.no	Types of trisomy	No. of cases	Percentage
1.	Primary trisomy 21 (47, XX or XY+21)	39	78
2.	Mosaic Trisomy 21 (46/47, XX or XY+21)	6	12
3.	Translocation 46, XY, t(14:21) 46, XY, t (21:21)	2	4
4.	Down's Syndrome Primary amenorrhoea (47, XX, +21)	3	6

Table 6: Classification of 50 down's syndrome cases according to the karyotype

**Fig. 1: Down's baby showing broad and short neck, flat occiput and excessive nuchal skin.**



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**Fig. 2: 2 year old Down's baby stands only on support.**



**Fig. 3: Down's baby showing scar mark of post-operative duodenal atresia and stenosis.**



**Fig. 4: Down's baby side profile showing opened mouth macroglossia, epicanthic fold, short and square nose oblique palpebral fissure and malformed ear.**



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**Fig. 5: Down's baby showing healed scar mark of repeated skin infection.**



**Fig. 6: Down's baby showing trunk and stumpy like limbs and increased gap between great toe and rest of the toes.**

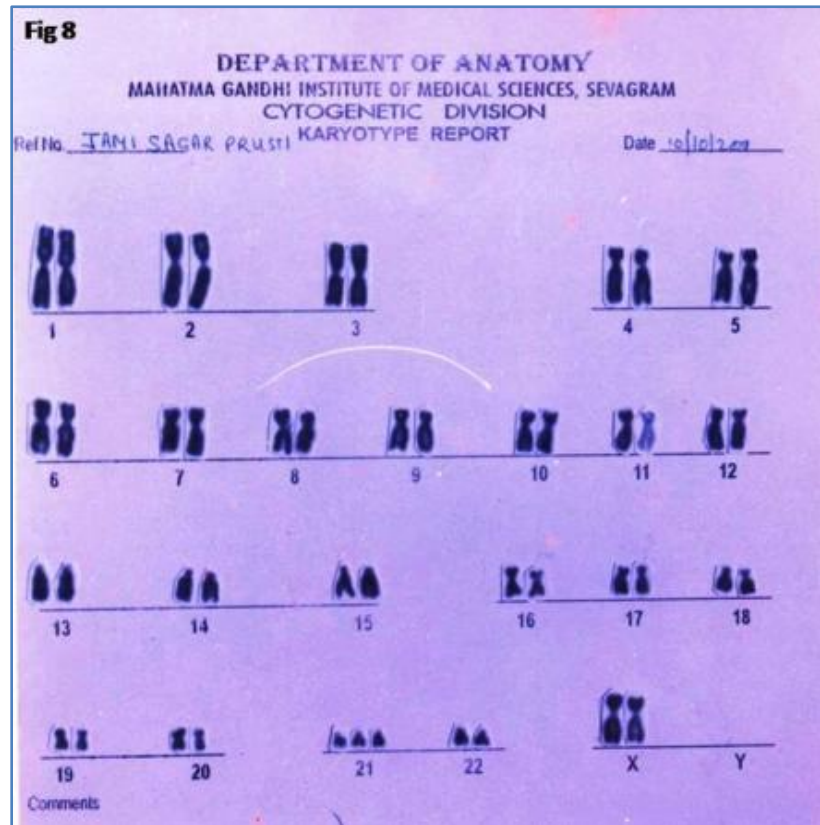


**Fig. 7: Measurement of height of a down's child by anthropometer.**



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**Fig. 8: Karyotype analysis of a Down's baby under high power microscope.**



### AUTHORS:

1. Jami Sagar Prusti
2. Sitansu K. Panda
3. Santosh K. Sahu
4. Dharma N. Mishra
5. Gitanjali Arora
6. Mahesh C. Sahu

### PARTICULARS OF CONTRIBUTORS:

1. Associate Professor, Department of Anatomy, MKCG Medical College, Berhampur, India.
2. Associate Professor, Department of Anatomy, IMS and SUM Hospital, Siksha O Anusandhan University, Bhubaneswar, India.
3. Assistant Professor, Department of Anatomy, SCB Medical College, Cuttack, India.
4. Assistant Professor, Department of Anatomy, VSS Medical College, Berhampur, India.
5. Associate Professor, Department of Anatomy, Hitech Medical College, Bhubaneswar, India.

6. Research Associate, Central Research Laboratory, IMS and SUM Hospital, Siksha O Anusandhan University, Bhubaneswar, India.

### NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Sitansu K. Panda,  
 Department of Anatomy,  
 IMS and SUM Hospital,  
 Siksha O Anusandhan University,  
 K8, Kalinga Nagar,  
 Bhubaneswar-751003, India.  
 Email: sitansupanda2011@gmail.com

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