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Exploring the Clinical Characteristics, Chromosomal Analysis, and Emotional and Social Considerations in Parents of Children with Down Syndrome

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Abstract:- Down syndrome, also known as trisomy 21, is a common chromosomal disorder that affects one in every 750/1000 live births. It is characterized by delayed milestones and repeated chest infections, with most children having microcephaly, low set ears, flat nasal bridge, simian crease, umbilical hernia, CVS findings, and repeated ear infections. The risk of Down syndrome increases to 1:20 for mothers over 40 years old. A study examined 30 children with phenotype suggestive of Down syndrome, with a male to female ratio of 0.76:1. The median age at presentation was 7 years, with a minimum of 3 years and a maximum of 12 years. The mean maternal age at child birth was 32 years, with 13.3% of the children born out of consanguineous marriage.

The study found that none of the mothers had a history of abortions, and 6.7% had complications during pregnancy. The most common complaint was delay in attainment of mile stones (96.7%) followed by recurrent LRTI & URTI (13.2%), with 2 having heart disease and 1 having family history of asthma.

Physical characteristics included microcephaly (86.7%), low set ears, a flat nasal bridge (96.7%), half of the children had simian crease and umbilical hernia (50%), excessive skin on the back (76.7%), muscular hypotonia (66.7%), dermatoses (26.7%), and hypothyroidism (10%). Cerebral findings were found in 15 children (50%), with endocardial cushion defects being the most common. ASD (13.04%) was most common in 12 children, followed by VSD (10.86%), and PDA (4.3%) was present in 4.3%.

Keywords: *- Down Syndrome, Abortions, Heart, Diseases, Hypothyroidism.*

I. INTRODUCTION

Down syndrome or trisomy 21 was first reported by Langdon Downin 1866. It is one of the most common chromosomal disorders occurring one in every 750/1000 live births. In mothers less than 25 years the risk is 1:2000, over 40 years the risk increases to 1:20.

All patients with Down syndrome have three copies of chromosome 21.95% have 47chromosome with trisomy of 21 chromosome.5% have 46 chromosomes with abnormally translocated 21st chromosome. Robertsonian translocation involves transfer of chromosomal materials from chromosome 21 to chromosome 13,14 or 15.

Having a child with mental retardation is considered a stigma in society and parents have to undergo a lot of mental and financial stress. Hence it was decided to take up the study to know the psychosocial aspects of parents as well as to correlate the clinical profile with genetics.

The study also analyses the knowledge the parents have, their perceptions about the condition, the issues after the diagnosis like the course, outcome, treatment options etc and the need to address the specific issues of individual families during counselling.

II. MATERIALS AND METHODS

Thirty children with phenotypic features of Down syndrome attending paediatric outpatient department of Government Wenlock Hospital and KMC Hospital, Attavar, Mangalore from November 2007 to August 2008, aged 3 to 14yrs were included in the study. Children were enrolled into the study as and when they approached either the OPD or IPD.

A written consent was taken after explaining the procedure to the parents Data concerning the clinical details were obtained from medical records of the study group.

During the visit, height was recorded on the stadiometer to the nearest 0.5 cm and weight on an electronic weighing machine to nearest 5 Gms. Head circumference was measured to the nearest millimeter using non stretchable fiberglass tape.

Microcephaly was defined as head circumference less than 3 standard deviation of mean for age. Blood pressure was measured in appropriate sized cuff covering 2/3 rd of arm circumference in upper limb.

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Blood samples were drawn and sent to genetic lab of KMC, Manipal for karyotyping for those children who had not undergone karyotyping before.

Psychosocial assessment of the parents was done with a predetermined semi structured pro forma.

The collected data was fed into the computer and analysis was done using SPSS version 11(Statistical Package for Social Sciences) statistical test chi square was used for analysing the qualitative data and for the quantitative variables student t test will be done. P< 0.05 will be taken as statistically significant.

III. RESULTS AND ANALYSIS

Table 1: Total Children Including in the study were 30, Out of which 13 are Male and 17 are Females

➢ *Sex*

	Frequency	Percent
M	13	43.3
F	17	56.7
Total	30	100.0

➢ *Age & Sex Distribution*

	Sex		Total
	M	F	
Less than 5 yrs Count %	9 47.4%	10 52.6%	19 100.0%
5-6 yrs Count %	4 44.4%	5 55.6%	9 100.0%
Gtr/eql Count %	0 0%	2 100.0%	2 100.0%
Total Count %	13 43.3%	17 56.7%	30 100.0%

Table 2: Consanguinity

	Frequency	Percent
2nd Degree Consanguinity	1	3.3
3rd Degree Consanguinity	3	10.0
Nil	26	86.7
Total	30	100.0

- 1 (3.3%) of children had history of second degree Consanguinity.
- 3 (10%) children had history of Consanguinity among Parents.

Table 3: The Number of Pregnancies in Mothers of the children with down syndrome 36% mothers were gravida 3 and 3.3% were Gravida 6.

Gravida

	Frequency	Percent
G1	2	6.7
G2	11	36.7
G3	9	30.0
G4	4	13.3
G5	3	10.0
G6	1	3.3
Total	30	100.0

Table 4: Frequency of Previous still birth and Abortion in the mothers 96.7% of mothers had no previous history of stillbirth and no history of abortion.

Still Birth

	Frequency	Percent
2	1	3.3
Nill	29	96.7
Total	30	100.0

Abortion

	Frequency	Percent
Nil	30	100.0

Table 5: Frequency of Previous Childbirth with Down Syndrome Only one Mother (3.3%) had a Child with Down Syndrome in Previous Birth

Previous Birth with Down

	Frequency	Percent
Nil	29	96.7
Normal	1	3.3
Total	30	100.0

Table 6: Type of Delivery in the Mothers 26 (86.7%) of the Mothers had Delivered Normally and 4 (13.3%) had Undergone LSCS

Delivery Type

	Frequency	Percent
LSCS	4	13.3
Normal	26	86.7
Total	30	100.0

Table 7: Complications during delivery 96.7% of mothers had no complications

Any Complications

	Frequency	Percent
Yes	1	3.3
No	29	96.7
Total	30	100.0

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Table 8: Complications in the Down Children after Delivery

Delayed Meconium

	Frequency	Percent
Yes	4	13.3
No	26	86.7
Total	30	100.0

Constipation

	Frequency	Percent
Yes	5	16.7
No	25	83.3
Total	30	100.0

Prolonged Jaundice

	Frequency	Percent
Yes	9	30.0
No	21	70.0
Total	30	100.0

Respiratory Distress

	Frequency	Percent
Yes	3	10.0
No	27	90.0
Total	30	100.0

Cardiac Complications

	Frequency	Percent
Yes	4	13.3
No	26	86.7
Total	30	100.0

Table 9: Chief Complaints in the children when presenting in the OPD 30% of the children presented to the OPD with complaints of delayed milestones 50% of the children had Umbilical hernia

Chief Complaints

	Frequency	Percent
No Complaints	14	46.7
Cold/Fever	2	6.7
Delayed Milestones	9	30.0
Development	1	3.3
Loose Stools & Vomiting	1	3.3
Recurrent Chest Infection	2	6.7
Recurrent cold / Chest	1	3.3
Infection	30	100.0
Total		

Umbilical Hernia

	Frequency	Percent
No	15	50.0
Yes	15	50.0
Total	30	100.0

Table 10: Clinical Features of Down Syndrome Observed in the children during presentation in the OPD

EYE

	yes	%	No	%
Upward Slanting	29	96.7	1	3.3
Brushfield spots	6	20	24	80
Anyother	28	93.3	2	6.7

ENT

	Yes	%	No	%
Ear Infection	6	20	24	80
Flat Nasal Bridge	29	96.7	1	3.3
Small Mouth	28	93.3	2	6.7
Protruding Tongue	22	73.3	8	26.7
High Arched Palate	27	90	3	10
Low set ears	29	96.7	1	3.3

- *Other Features Observed in the Down Children*

Skin Dermatoses

	Frequency	Percent
No	22	73.3
Yes	8	26.7
Total	30	100.0

Excessive Skin on the Back

	Frequency	Percent
No	9	30.0
Yes	21	70.0
Total	30	100.0

Muscular Hypotonia

	Frequency	Percent
No	10	33.3
Yes	20	66.7
Total	30	100.0

Hyper Flexibility

	Frequency	Percent
No	7	23.3
Yes	23	76.7
Total	30	100.0

Simian Crease

	Frequency	Percent
No	15	50.0
Yes	15	50.0
Total	30	100.0

Hypothyroidism on Treatment

	Frequency	Percent
No	27	90.0
Yes	3	10.0
Total	30	100.0

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Microcephaly

	Frequency	Percent
Microcephaly	26	86.7
Normal	4	13.3
Total	30	100.0

Table 11 Karyotyping Results

Report of Chromosome analysis and Karyotype

- Patient Name: Abhay Krishna
- Case Name: L003361
- Age: 6 yrs
- Date of Birth: 8/03/08
- Specimen Type: Peripheral Blood
- Referral Reason: D.S
- Result: Chroosome analysis by blood Microculture and GTG branding shows 47, XY, + 21 in all the 20 metaphase plates analysed.
- Case Comment: 1 Cytogenetically this is a case of free trisomy 21 indicating Down syndrome. 2 Genetic counselling advised. 3. Prenatal diagnosis can be offered in cause of future pregnancies.

Karyotyping

	Frequency	Percent
Mosaicism Robertsonian	1	3.3
Translocation	1	3.3
Trisomy 21	28	93.3
Total	30	100.0

Table 12: Showing Details of Psychosocial Issues in Parents of the Down's Children

Mothers Occupation

	Frequency	Percent
Homemaker	27	90.0
Tailor	1	3.3
Teacher	2	6.6
Total	30	100.0

Mothers Education

	Frequency	Percent
Graduate	5	16.6
PUC	15	50.0
SSLC	10	33.3
Total	30	100.0

How Parents Came to know the Condition of the Child

	Frequency	Percent
Appearance of Child	14	46.7
She was not able to	12	40.0
sit/crawl.	4	13.3
Doctor diagnosed.	30	100.0
Total		

Knowledge about cause of the condition

	Frequency	Percent
Excess of work	2	6.7
during Pregnancy	2	6.7
A curse of the Gods	26	86.7
A Medical Cause	30	100.0
Total		

Reaction to the Diagnosis

	Frequency	Percent
Depressed	15	50.0
Felt Guilty	5	16.7
Fraustrated/Worried	5	16.7
Acceptance	5	16.7
Total	30	100.0

Source of Help approached

	Frequency	Percent
Doctor	25	83.3
Faith Healer	3	10.0
Counselor	0	0
Religious Leader	2	6.7
Total	30	100.0

Support for the Family and Child

	Frequency	Percent
Family Support	25	83.0
Emotional & Financial	0	0
No, No Support from	5	17
family members	0	0
Support from social	30	100.0
Organisations		
Any other		
Total		

Telling the family about the condition and needs of children

	Frequency	Percent
Have Told	24	80.0
Haven't Told	2	7.0
Have told, but	4	13
interpersonal		
Relationship	30	100.0
affected		
Total		

Relationship of the Child with Siblings

	Frequency	Percent
Patient and caring towards	18	60.00
the child	2	6.70
Don't involve in the	10	33.30
activities of the child	0	00
Get frustrated at times	30	100.0
Make fun of the child		
total		

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Time Spent by Child during the Day

	Frequency	Percent
At home	20	66.7
Special School	10	33.3
Normal School	0	0
Others	0	0
Total	30	100.0

Frequent Change of School

	Frequency	Percent
Cannot learn or play	0	0
Other children do not join him in play	0	0
Doesn't change schools often	11	37.3
Stays at home	19	63.3
Total	30	100.0

Does Parent Get Time for Leisure Activities

	Frequency	Percent
Most of time is spent with child	9	30.0
Get enough time for self	16	53.0
Gets time and involves child too in recreational activities	5	17
Total	30	100.0

Situation at home when there are visitors

	Frequency	Percent
Child kept away from visitors	25	83.3
Visitors don't interact with child	2	6.7
Child involved in interaction with visitors	3	10
Total	30	100.0

Family Participation in Socio Religious Functions/Family Gatherings?

	Frequency	Percent
Participate with the child	25	83.3
Participate without the child	2	6.7
Don't participate in family functions	3	10
Total	30	100.0

Relationship of child with neighbours

	Frequency	Percent
No Interaction with Child	5	16.70
Problems in the neighbourhood	23	76.70
Children in neighbourhood play with child	0	00
Total	2	6.70
	30	100.0

Involvement in Child's School Activities

	Frequency	Percent
Visit School and PTA	10	33.30
Help with homework and teach at home	8	26.70
Don't involve with school activities	12	40.00
Total	30	100.00

Letting child play with other children in the neighbourhood

	Frequency	Percent
Often	11	36.70
Sometimes	18	60.00
Don't let child Play	1	3.30
Total	30	100.0

Special Quality do Recognised in Child

	Frequency	Percent
Music and dance	21	70.00
Drawing using colors	2	6.70
Both	7	23.30
Total	30	100.0

Encouraging Child to Participate in Games and Sports

	Frequency	Percent
No	10	33.30
Yes	4	13.30
Cant say	16	53.30
Total	30	100.0

Worry about Future of Child When He/She Grows Up

	Frequency	Percent
Child can never be independent	10	33.30
Child will be able to earn his living on his own	9	30.00
Cant say	11	36.70
Total	30	100.00

Ways Tried to Make Child Confident like any other Normal Child

	Frequency	Percent
Encourage to speak to people/Take child our with the family	11	36.70
Make child feel like a normal child	8	26.70
Unable to instill confidence in child	11	36.70
Total	30	100.0

Leaving the Child by Himself

	Frequency	Percent
Yes, Most of the time	2	6.7
Feel Embrassed when child gets into trouble with others	2	6.7
Afraid child will hurt himself/others will hurt him	26	86.7
Total	30	100.0

Getting Annoyed and Impatient when Teaching Child

	Frequency	Percent
Often	0	00
Sometimes	14	46.70
Don't get annoyed	16	53.30
Total	30	100.00

Attempt Made to Improve Vocabulary of Child

	Frequency	Percent
Speak to child and explain	3	10.0
Child able to understand & Speaks day to day things	10	33.3
Difficuly to speak as child cannot	17	56.7

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understand		
Total	30	100.0

The Risk Child Faces the Most

	Frequency	Percent
Social Rejection and Loneliness	11	36.7
Scholastic Backwardness and	18	60.0
Dependence	1	3.3
Child Abuse	30	100.0
Total		

Monthly Expenditure on the Child (in Rupees)

	Frequency	Percent
Greater 500	0	0
500-1000	21	70.0
Less 1000	9	30.0
Total	30	100.0

Bearing the Expenses of Child

	Frequency	Percent
Parents	24	80.0
Voluntary	6	20.0
organisation	0	0
Government	30	100.0
Total		

Professional Counselling Being Done for the Child

	Frequency	Percent
No	9	30.0
Monthly	3	10.0
Only When need	18	60.0
arises	30	100.0
Total		

Have the Parents Enrolled in any Support Group

	Frequency	Percent
Yes	5	16.7
No	25	83.3
Total	30	100.0

IV. DISCUSSION

Down syndrome is a common autosomal abnormality in humans, affecting 1 in 750 live births. In this study, 30 children with phenotype suggestive of Down syndrome were studied, with a male to female ratio of 0.76:1. The median age at presentation was 7 years, with a minimum of 3 years and a maximum of 12 years. The mean maternal age at child birth was 32 years, with 13.3% of the children born out of consanguineous marriage.

The study found that none of the mothers had a history of abortions, and 6.7% had complications during pregnancy. The most common complaint was delay in attainment of mile stones (96.7%) followed by recurrent LRTI & URTI (13.2%), with 2 having heart disease and 1 having family history of asthma.

Physical characteristics included microcephaly (86.7%), low set ears and a flat nasal bridge (96.7%). Half of the children had simian crease and umbilical hernia (50%), excessive skin on the back (76.7%), muscular hypotonia (66.7%), dermatoses (26.7%), and hypothyroidism (10%).

Cerebral findings were found in 15 children (50%), with endocardial cushion defects being the most common. ASD (13.04%) was most common in 12 children, followed by VSD (10.86%), and PDA (4.3%) was present in 4.3%. Mental IQ assessment revealed that 18 out of 92 children had mild mental retardation (IQ 50-70), most were in the moderate group (IQ ranging from 35 to 50), 60 children had moderate mental retardation, and 14 children had severe mental retardation (IQ 20-35).

The study reveals that 93.3% of children with Down syndrome have trisomy 21 with non dysjunction, while 3.3% have Robertsonian translocation and mosaicism. Out of the 30 children, 90% are homemakers, 33.3% are educated up to the tenth standard, 50% are graduates, and 10% are employed. Parents come to know their child's condition by appearance, 40% by delayed milestones, and 13.3% are diagnosed by a doctor. 86.7% of parents are aware of the condition, while 6.7% believe it is caused by excess work during pregnancy.

50% of parents are depressed when they know their child has Down syndrome, while 6.7% feel guilty and 6.7% accept the condition. 83.3% of parents approach a doctor, 10% seek a faith healer, and 6.7% go to a religious leader when they first learn about the condition. 83% of parents have support from family members and 17% have support from social organizations.

80% of parents have told their family members about the condition, while 7% have not. In 13% of cases, the interpersonal relationships of family members have been affected. Siblings are patient and caring towards the Down child, while 61% get frustrated and 6.7% do not involve in any of the child's activities.

66.7% of Down's children spend most of the day at home, and 33.3% attend special school. Parents have enough time for leisure activities, and 83.3% participate in family functions with the child. Visitors who come home do not interact with the child.

76.7% of children face problems in the neighborhood, and 16.7% have no interaction with the child. 40% of parents do not involve in school activities, while the rest participate in PTA meets and help with homework.

53.3% of parents teach the child basic skills like brushing and bathing, and 33.3% teach them small moral lessons. Only 60% of parents let their child play with other children in the neighborhood.

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Most parents believe that their child will face academic backwardness and dependance, social rejection and loneliness, and child abuse. Monthly expenses for the child range from 500-1000 rupees, with 80% of parents bearing the costs themselves.

V. CONCLUSIONS

The study examined 30 children with Down syndrome, focusing on their clinical features, karyotyping results, and psychosocial issues. The most common complaint was delayed milestones and repeated chest infections. Most children had microcephaly, low set ears, flat nasal bridge, simian crease, umbilical hernia, CVS findings, and repeated ear infections. Karyotyping results showed that 93.3% of the children had trisomy 21 with non dysjunction, Robertsonian translocation, and mosaicism, making trisomy 21 nondysjunction the most common chromosomal anomaly among the three variants.

Parents' psychosocial issues depend on their knowledge about the condition, family relationships, parental adjustment to the child's demands, financial status, facilities available for the child, and support from extended family and social groups. Parents who were well-informed about Down syndrome showed improved adjustment, less stress, and a more positive perception of their child. Quality of life for the child was better in families where members felt connected and supported by each other.

Sibling relationships were also crucial for children with Down syndrome. Most children faced neighborhood problems and less interaction with visitors, but parents played a significant role in their child's development and family functioning. Many parents taught basic skills like brushing, bathing, and moral lessons, and recognized their interest in music and dance.

Approximately 80% of parents were not involved in any support group and only sought counseling when needed. There was less awareness about services and support available to children with Down syndrome. Early intervention programs, pre-school nurseries, and inclusive special education strategies positively influenced children's overall functioning.

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