

Issues in Counseling for Down Syndrome

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Manuscript received: January 9, 2006; Initial review completed: May 1, 2006;

Revision accepted: August 2, 2006

We studied the background information, concerns and specific queries of thirty-four families of children with Down syndrome. Majority of the parents were aware that their child has Down syndrome and has or will have mental retardation. However, most of the families were ignorant about the lack of curative treatment, chromosomal nature of the disorder and prenatal screening and testing options.

Key words: *Counselling, Down syndrome.*

With an incidence of 1:700 to 1:1000, Down syndrome is the most common cause of referral for genetic counseling(1-3). As there is no curative therapy, genetic counseling has a central role to play in the management. Counseling for Down syndrome is complex and each case is challenging for even an experienced counselor. A good counselor has to be alert to the background awareness of parents and caretakers as well as be prepared for their queries, some of which erupt spontaneously during the process of counseling. The present study was planned to analyze the background information the parents have and document the issues that arise during counseling.

Parents of children with Down syndrome attending medical genetics outpatient department of a tertiary referral centre over a period of one year were enrolled for the study and counseled in accordance with the guiding principles(4). The counselor documented the background information that the consultands had about Down syndrome with the aid of a predetermined protocol. The proforma included information regarding awareness of: Down syndrome in their child, presence of mental retardation, incurability of the condition, inability to formal schooling, different look from other children, chromosomal etiology of the condition, trisomy 21,

relation to maternal age, feasibility of prenatal screening and diagnosis and other relevant information. The major concerns of the parents and questions arising during the course of interview and counseling as perceived by the investigator were also recorded.

Thirty-four patients (2 neonates, 19 infants and 13 older children) and their families were studied. Of these, nine were diagnosed to have Down syndrome in the neonatal period, 19 during 1-12 months of age and 6 at a later age. Twenty-five children were referred by the pediatricians. The influences of age at the time of presentation and educational status of parents on background information are shown in *Table I*. The higher of the qualification of the parents was considered for interpretation of the results. Major concerns noted by the parents were mental retardation(21), schooling(12), cardiac problems(6), recurrence of the condition(6), communication(5), intercurrent problems(5), treatment(3) and reproductive ability(3).

Some notable queries those arose during the counseling and the beliefs the parents had are: will others recognize him as mentally retarded child by his look?; whose defect is it (father's/mother's)?;

TABLE I—Influence of Age at Presentation and Educational Status of Parents on Background Information

Information	Awareness if presentation during infancy (n = 21)	Awareness if presentation beyond 1 year (n = 13)	Awareness if At least one graduate parent (n = 22) (Median age at presentation 9 months)	Awareness if neither is a graduate (n = 12) (Median age at presentation 1.9 years)	Total (n = 34)	
					Aware	Not aware
Down syndrome in their child	15	9	18	6	24	10
Mental retardation in this condition	14	11	20	5	25	9
Lack of curative treatment	4	5	8	1	9	25
Different look from others/ facial dysmorphism	11	8	13	6	19	15
Inability to have formal schooling	4	3	5	2	7	27
Chromosomes	9	2	9	2	11	23
Trisomy 21 in their child	1	1	2	0	2	32
Relation with maternal age	2	3	3	2	5	29
Feasibility of prenatal screening and diagnosis	4	1	5	0	5	29

will he earn his livelihood?; how to kill this child (as it is worthless for him to live)?; were told that he will be able to study in Hindi medium; We are told that he/she will not survive beyond 17-18 years; Down syndrome occurs due to improper antenatal care; he will be mad; is it a lethal condition?; heart defect need not be operated in children with Down syndrome; do they have reduced life expectancy?; will his/her children also have Down syndrome?; screening is done only after the first child with Down syndrome; happy to note that the child will not have major physical problems.

The findings of our study show that many parents are not well informed about the condition including that their child has Down syndrome. This could be due to several reasons. Difficulties in diagnosis of a neonate with Down syndrome or unwillingness on

the part of the pediatricians to break the bad news are possible explanations. It is also obvious from the study that the families are rarely informed about the antenatal screening and testing options, at least for the subsequent pregnancies. This finding demands a serious consideration in educating the people about preventive strategies including routine use of triple test.

Several unique concerns/views were expressed by few families. It is an integral part of the care of these children to understand how the family feels and reacts to the facial appearance. Few families were worried about being pointed out in the society whereas one family felt their son looks handsome! Some of them verbalized specifically about common aspects like education, job and longevity. In an Indian set up, several families affected with

What this Study Adds

- Families with a child with Down syndrome are poorly informed of the condition and physicians need to address the specific issues of individual families during counseling.

incurable conditions also raise issue of marriage and reproductive ability. The counselor should be cautious and at the same time be truthful while approaching these kind of issues. The counselor needs to find out the perceptions of the family about the condition and burning issues after the diagnosis like course, outcome, treatment options, cause, blame etc. These deserve special emphasis for a given family while giving them realistic picture of the future.

Contributions: SRP: Concept, design, critical revision of the draft; GKM: Collection, analysis of data, drafting of the manuscript; SVS: Analysis of data

Funding: None.

Competing interests: None stated.

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Seroprevalence of *Chlamydia pneumoniae* in Asthmatic Children from Northern India

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Manuscript received: August 18, 2005; Initial review completed: December 15, 2005;

Revision accepted: October 9, 2006.

Serum from children with chronic persistent asthma was subjected to C. pneumoniae IgG antibody determination. C. pneumoniae IgG serology suggestive of persistent infection was significantly higher in chronic persistent asthma group than in the control group. Seropositivity was significantly more in moderate and severe persistent groups than in the control subjects. There was no evidence of acute C. pneumoniae infection (IgM serology done in duplicate) in acute exacerbations.

Key words: *Chlamydia pneumoniae, Chronic asthma, Serology.*

C. pneumoniae has been shown to infect human smooth muscle cells(1), generate inflammatory cytokines in vitro(2) and has also been linked to asthma. Most of the studies evaluating association between *C. pneumoniae* and asthma have been done

in adults(3-5). Information in children is scarce with most reports showing association with acute exacerbations of asthma(6-8). We evaluated the potential role of *C. pneumoniae* infection in chronic persistent asthma and acute exacerbation of asthma.