

Psychiatric Evaluation of Children with Fragile X-syndrome and Down Syndrome

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ABSTRACT

Background: Down syndrome (Trisomy 21) is one of the commonest syndromes with psychiatric aspects. Fragile-X syndrome is also known chromosomal disorder with neuron-psychiatric disorders. **Objective:** to identify neuropsychiatric problems commonly found in Down syndrome and Fragile-X syndrome. This has impact on both understanding neuropsychology of common behavior problems in children and in their management. **Subject and methods:** They were recruited from national Center for Research they were 15 children with Down syndrome and 15 children with Fragile-X syndrome. We used the clinical condition checklist and childhood Autism rating scale. **Results:** There was high incidence of low IQ among the 2 groups, increased hyperactivity and inattention in both groups. **Conclusion:** Children with Fragile-X syndrome showed high incidence of autistic disorder. Discussion of the results was done and recommendations were given for further researches.

Key words: Fragile X- syndrome, Down syndrome, Psychiatry

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INTRODUCTION

Behavior phenotypes are the characteristic cognitive, personality, behavior and psychiatric patterns that typifies a disorder¹, some patients will be referred because of their behavior problems to identify the presenting symptoms as apart of behavioral phenotype will allow referral to a geneticist and accurate genetic diagnosis. This is important for parental counseling regarding prognosis and risk in the following pregnancies. It also allow early interventional and treatment recommendations².

Down syndrome (Trisomy 21) is one of the commonest syndromes with psychiatric

aspects. It has incidence of 1 in 600 live births³. About 20% to 40% of children with Down syndrome have behavior problems such as aggression and hyperactivity⁴. These problems persist through adulthood⁵.

Fragile-X syndrome is also known chromosomal disorder with neuron-psychiatric disorders. It is caused by mutations in a single gene in the left arm of the X chromosomes (X-27.3). It occurs in 1/2000-4000 live births in males and 1/6000-8000 in females. Affected males show specific physical features in the form of broad forehead, elongated face, large prominent ears and high arched Palat⁶.

Children with Fragile-X syndrome suffer from mental retardation in the mild-severe range². In addition children with Fragile X syndrome display a characteristic profile of behavior disturbances⁷ includes social deficits with peers, abnormalities in communications, stereo-typed behavior, impulsivity and hyperactivity.

The aim of this work is to recognize in systematic and accurate way the behavior and psychiatric syndromes that may be associated with both Down syndrome and Fragile-X syndrome.

This will help us in giving the prone care in all aspects of these handicapped children.

SUBJECTS AND METHODS

Thirty patients were collected from the National Research (clinic of children with special needs), 15 children was diagnosed as Down syndrome and 15 as Fragile-X syndrome.

Inclusion criteria for selection were the laboratory diagnosis of chromosomal abnormalities and the age group of the selected cases ranged from 4-17 years, so we have two groups:

Group I (N=15) includes children with Down syndrome (7 males and 8 females). Group II (N=15) includes children with

Fragile-X syndrome. All of them are of course males.

The two groups were subjected to Stanford Binet Intelligence Test⁸, test for Assessment of the ADHD and its severity Conner's Parent Rating Scale –Revised long version (CPRS-R-L)⁹, Children Behavior Checklist for ages 4–18 (CBCL 4 – 18)¹⁰ and test for Diagnosis and Rating the Degree of autism: Childhood Autism Rating Scale (CARS)¹¹.

RESULTS

The results of our work showed four aspects of abilities and behavior problems of two important chromosomal syndromes. The major ability which is the intelligence measured by Stanford Binet (table 1) showed that children with Down syndrome lie between mild and moderate IQ each of them 50% of the group. On the other hand only 20% of children with Fragile-X syndrome show Borderline IQ while 40% in the mild degree and the rest was in moderate and severe degree.

Autistic disorder associated with Fragile-X syndrome was significantly evident in this study as shown in table (2) compared to association with Down syndrome.

Table (3) shows comparison between both groups CBCL. Both groups showed higher scores than norms on this test but no differences between both groups.

Table (1) IQ assessments of the groups

IQ	Group 1 (N= 15)	Group 2 (N= 15)	P
	Down syndrome	Fragile -X	
Dull average 80-90	0	0	NS
Borderline 70-80	0	3	0.05
Mild MR 55-70	8	6	NS
Moderate MR 40-50	7	4	NS
Severe MR 25 -40	0	2	NS

Table (2): Comparison between Down syndrome & Fragile-X syndrome regarding Autism

CARS	Group 1 (N= 15)	Group 2 (N= 15)	P
	Down syndrome	Fragile -X	
Non Autistic	15	5	0.001
Mild to Moderate Autism	0	7	0.001
Severe Autism	0	3	0.001

Table (3) comparison between Down syndrome & Fragile-X syndrome on CBCL

CPCL	Group 1 (N= 15)	Group 2 (N= 15)	P
	Down syndrome	Fragile -X	
Social competence Mean $\pm$ SD	39.1 $\pm$ 9.6	33 $\pm$ 7.8	>.05 NS
Total competence Mean $\pm$ SD	31 $\pm$ 5.6	28.4 $\pm$ 6.2	>.05 NS

Table (4) comparison between Down syndrome and Fragile-X syndrome on Hyperactivity

	Group 1 (N= 15)	Group 2 (N= 15)	P
	Down syndrome	Fragile -X	
Hyperactivity Subscales	Mean = 58 $\pm$ 14	Mean = 69 $\pm$ 16	0.06 NS
• normal range	8	3	
• borderline	2	2	
• Possible problems	0	3	
• Evident problem	1	0	
• Significant problem	4	7	
Restless Impulsivity	Mean = 59 $\pm$ 9.8	Mean = 62 $\pm$ 10	0.32 NS
• Normal range	5	4	
• Borderline	4	2	
• Possible problem	2	1	
• Evident problem	2	4	
• Significant problem	2	4	
DSM-IV Hyperactivity	Mean = 61.8 $\pm$ 13	Mean = 66.7 $\pm$ 14	0.36 NS
• Normal range	6	3	
• Borderline	2	1	
• Possible problem	2	1	
• Evident problem	1	3	
• Significant problem	4	1	

DISCUSSION

Genetic disorders have specific effect on behavior. This abnormal behavior is termed behavior phenotype of the syndrome.

In our research we selected two well-known syndromes, the Down syndrome and Fragile-X syndrome for deep systematized study.

As regard patients with Down syndrome assessment of intelligence revealed they suffer from moderate MR. this result is in agreement with previous studies done by¹²⁻¹⁴ who reported that Down syndrome patients have mild to moderate cognitive impairment cognitive abnormalities in Down syndrome is caused by brain abnormalities cortical lamination and irregular clustering of neuronal cell bodies¹⁵.

Social problems are associated in some cases of Down syndrome are yet within borderline clinical range. These results are in agreement previous results which concluded that socialization in Down syndrome was mainly through non-verbal modalities as they had weakness in communication relative to daily living and socialization skills¹⁶.

On the other hand and against our results the diagnosis of conduct disorder was detected in a considerable ratio in children with down syndrome¹⁷⁻¹⁸.

Another finding in agreement with our study showed that patients with Down syndrome have hyperactivity and attention problems¹⁷.

As regard patients with Fragile-X syndrome assessment of intelligence revealed that affected children fall in the range from mild to severe MR. These results are in agreement with previous studies that experienced this cognitive disabilities by

minor structural brain abnormalities found in Fragile-X syndrome¹⁹⁻²⁰. Also individuals with Fragile-X syndrome were put within the range of moderate MR⁷. Another study found that individuals with Fragile-X syndrome has no MR but only dull normal intelligence²¹. There is evidence from another longitudinal study that there was progressive decline in intellectual abilities with age in individuals with Fragile-X syndrome²².

Autistic symptoms were present in most cases with Fragile-X syndrome ranging from mild to moderate as previously reported²³⁻²⁴.

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