Restricted Repetitive Behaviors Phenotype? Calling Attention to Children Who Are Not Diagnosed with ASD and Exhibit Excessive Interest in Specific Topics or Repetitive Behaviors

Abstract. The aim of this report is to call attention to a unique clinical group of children who are not diagnosed with ASD but exhibit excessive interest in specific topics or repetitive behaviors. It is argued that this group might require specific developmental stimulation, especially in light of the fact that it does not receive adequate professional treatment or support. The results of conducted surveys are supplemented with additional diagnostic and anamnestic data from which practical conclusions based on research evidence are drawn and general guidelines for intervention are formulated. Further research into this group is also suggested.

Keywords: ASD (autism spectrum disorder), DSM-V (Diagnostic and Statistical Manual of Mental Disorders), ADOS (Autism Diagnosis Observation Schedule)

Słowa kluczowe: ASD (Spektrum zaburzeń autystycznych), DSM-V, ADOS, (Protokół obserwacji do diagnozowania zaburzeń ze spektrum autyzmu)
INTRODUCTION

The aim of this report is to call attention to a unique clinical group of children, who are not diagnosed with ASD and exhibit excessive interest in specific topics or repetitive behaviors. The children we encounter often have relatively preserved interactive conversational skills and suitable non-verbal communication (e.g., eye contact, facial expressions, and hand gestures), while having social difficulties with same-age peers but not with adults, and show excessive interest in unusual or highly specific topics/objects or repetitive behaviors. These children often have linguistic difficulties including word-finding difficulties and poor vocabulary. Our mission is to call for professional attention to this clinical group which does not receive adequate support and treatment, and stimulate further research into this group. This important finding is triangulated with additional diagnostic and anamnestic data, and general guidelines for intervention are suggested.

BROADER AUTISM PHENOTYPE

The Broader Autism Phenotype (BAP) refers to different constellations of autistic traits usually examined in the context of sub-diagnostic phenomena in families of individuals with Autism Spectrum Disorder (ASD) (e.g., Pisula, Ziegart-Sadowska, 2015). Our focus in referring to BAP is different. We examine different constellations of autistic traits, namely, restricted and repetitive behaviors and interests (RRBs) and social communication, to describe a sub-diagnostic group that warrants clinical attention.

RRBs, together with social-communication difficulties, constitute the main domains of ASD (American Psychiatric Association [APA], 2013). Harrop et al. (2014) compared toddlers diagnosed with ASD to typically developing (TD) ones, and found that although RRBs were much more prevalent among children with ASD, among the TD children, RRBs were generally limited to one form of behavior (“fiddles with objects / uses objects in repetitive and non-functional manner”). Harrop et al. (2014) proposed that the developmental function of RRBs may be different for the two groups; TD children may exhibit these RRBs in order to learn how the toys work and master their use, while children with ASD have difficulty in shifting attention, and use perseveration with a non-functional play to serve self-stimulatory purposes. Harrop et al. (2014) found that in the ASD group, RRBs were negatively correlated with the total language score (expressive and receptive combined), while this association did not reach significance in typically developing children.

Troyb et al. (2014) found that some of the children, who had been diagnosed with ASD in the preschool period, achieved optimal outcomes years later; i.e., no longer met diagnostic criteria for ASD. Since symptoms of RRBs tend to persist into adolescence and adulthood, even when there is an improvement in social and communication symptoms of ASD, Troyb et al. (2014) claimed that individuals who had RRBs symptoms were less likely to undergo “recovery”, and thus it is possible that RRBs are the ASD domain that is least likely to improve over time. Therefore, RRBs may persist among children whose diagnosis was removed at an older age. Children, who display RRBs during preschool years, have been shown to have a tendency to present poor linguistic abilities and outcomes (Charman et al., 2005; Paul et al., 2008).

METHOD

Participants

This research focuses on a group of 38 boys who were referred for an ASD diagnostic assessment, and were found to have a preponderance of excessive interest in specific topics or repetitive behaviors, while their overall profile was sub-clinical for ASD. We compared them to 40 boys who were diagnosed with ASD the same year. We controlled for gender by removing from our sample two girls who were diagnosed that year. There was no significant difference between the mean age of the groups: the mean age in the group of boys diagnosed with ASD was 10.43 (SD = 2.80) and the mean
age in the other group was 9.77 ($SD = 2.65$); $t_{76} = .28$, n.s. A large proportion of the children (40.3%) used psychotropic medications, and 72.8% of the parents lived together while the rest were either single mothers or divorced. In this comparison, we controlled for intelligence by including only boys with IQs over 70 who were referred for ASD diagnosis.

**Materials and Design**

As part of our service as the autism center in a public hospital, we provide diagnostic assessments focusing on ASD to children ages 6–18 from all over Israel. Children are referred by neurologists, psychiatrists, psychologists, and other assisting professionals, because they have demonstrated social communication difficulties and sometimes social exclusion. The assessment is comprehensive, multidisciplinary and includes the use of the Autism Diagnostic Observation Schedule – Second Edition (ADOS-2; Lord et al., 2012), Childhood Autism Spectrum Test (CAST; Scott et al., 2002), a teacher questionnaire, a parent questionnaire, an anamnestic interview with both parents, and a structured interview based on the DSM-5 criteria. Our assessment procedure mandates that each evaluation be conducted by a team of experienced and senior neurologists, psychiatrists, and psychologists.

The clinicians described myriad difficulties that the RRB participants displayed. The descriptions of RRBs included “repetitive movements”, “being preoccupied with food”, “difficulties in self-regulating”, and “restlessness”. The social difficulties included describing participants as “lonely”, “without any friends”, “always alone”, and “lack of interaction”. Academic difficulties were described as “learning difficulties”, “difficulty adapting to school”, and “being completely dependent on teacher mediation”. Linguistic difficulties included: “having difficulty expressing himself”, “treated by speech and language pathologist from the age of two until today”, and “using language in idiosyncratic ways”. Finally, other difficulties included “behavior problems”, “anxieties”, and “anger”.

**Procedures**

After obtaining the necessary permissions from the hospital’s ethical review board, all children whose score on the ADOS-2 D4 item was greater than 0 and had not received a diagnosis of ASD were collected. This procedure led to the formation of the “RRB group” comprising 38 boys, who were then compared to a group composed of 40 boys who were diagnosed with ASD (henceforth “ASD group”). The data was analyzed qualitatively and quantitatively. The qualitative descriptions given by the clinicians’ interview notes, as well as the parent and teacher questionnaires, were quantified and added to the quantitative data from the standardized measures. The data was analyzed using SPSS software to produce descriptive statistics, and to compare the RRB and the ASD groups.

**RESULTS**

The findings indicated the following characteristics of the RRB group:

1. The RRB group had a significantly lower mean score (indicating fewer difficulties) on the Social Affect Domain of the ADOS-2 ($F = 17.113; p = .000$).

2. A high percentage of children presented behaviors described in item D4 in ADOS-2: “Excessive interest in unusual or highly specific topics/objects or repetitive behaviors”. Figure 1 presents the distribution of D4 scores among the two groups. It illustrates the differences in the ratio of repetitiveness and excessive interest between the ASD and the RRB groups: about 30% of the boys in the ASD group presented RRBs compared with 52.6% in the RRB group.

3. There was a preponderance of linguistic difficulties in the RRB group. In 60% of the cases the diagnostician noted naming difficulties, retrieval difficulties and/or poor vocabulary.

4. We explored demographic variables that may be related to the RRB group, and three background variables were found to be sig-
significant: school type (special/regular), parental marital status (married/non-married), and use of medication (yes/no).

a) School type: a Pearson $\chi^2$ test indicated a significant association between school type and ASD diagnosis. Children who attended special schools were more likely to be diagnosed with ASD ($\chi^2 = 5.419; p = .020$) (see Table 1).

b) Parental marital status: a two-way ANOVA on CAST scores revealed a significant interaction effect between parental marital status and the type of group (i.e., ASD versus RRB). While no differences have been found between married and non-married parents with regard to the CAST score in the ASD group, there is a significant interaction effect whereby non-married parents reported a higher CAST score than their married counterparts in the RRB group ($F_{1, 76} = 4.995; p = .029$).

c) Use of medication: a Pearson $\chi^2$ test indicated no significant association between use of medication and being diagnosed with ASD ($\chi^2 = .002; p = .961$) (see Table 2).

**DISCUSSION**

The RRB group in this study was subclinical for ASD while exhibiting RRBs, linguistic and social difficulties, and having been prescribed medication to the same extent as the ASD group. These are significant findings indicating the extensive clinical attention that this group of children requires despite not having a formal diagnosis. Children in the RRB group did not elicit as much educational attention as children in the ASD group with regards to placement in special education. We propose that cases of RRBs with no overall diagnosis of ASD should be further examined and studied to determine whether this clinical group represents a unique disorder. Tentatively, we suggest naming this disorder “RRB phenotype”. Children who belong to the RRB phenotype group are characterized by repetitive and restricted behaviors, excessive interest in unusual topics or objects, as evaluated by the ADOS-2. While there may be social difficulties, they are not primary but secondary, not as significant as those of ASD, and they may be also qualitatively different. Our

![Figure 1](image-url.png)

**Figure 1.** Distribution of D4 code scores on among ASD and RRB groups

Source: own elaboration.
findings and clinical experience indicate that these children have difficulties in the academic and social domain that warrant clinical attention. We suggest that RRBs both alienate them from their peers, as well as provide refuge from the intense emotions that accompany social interactions, thus hindering their social skills. It is also possible that comorbid linguistic difficulties are at the root of the children’s social difficulties. Finally, another developmental trajectory that should be further explored in future research is whether there are children who “grew out of autism” thanks to intensive early intervention, and only the RRB symptoms remained, as previous research suggests that these symptoms are the most resilient (Troyb et al., 2014).

**Implications**

The study has implications with regards to assessment practices. In particular, we found that CAST scores were higher for non-married rather than married parents. This may be due to the excessive stress of divorce or single parenting, and diagnosticians should be made aware of this bias.

We recommend the following therapeutic intervention to individuals characterized by RRB phenotype. First and foremost, professionals should be made aware of the possible existence of the clinical group as characterized here. Many interventions may be highly relevant to RRB phenotype children including focused cognitive behavioral therapy (Wood et al., 2021), social-skills training (Wolstencroft et al., 2018), and speech and language therapy. Interventions should target the inclusion of children with RRBs in society, helping them function as best as possible, and improving their quality of life.

We suggest that these children often fall through the cracks, and are not provided enough support because they do not have the ASD diagnosis. These children often need assistance, which is hard to come by without a diagnostic label. Along with the significance

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**Table 1.** Type of school attended by boys diagnosed with ASD versus boys not diagnosed with ASD

| Groups | Regular school |  |
|--------|---------------|------|
|        | No | %    | Yes | % |
| N      |    |      | N   | % |
| RRB    | 13 | 28.9 | 27  | 71.1 |
| ASD    | 22 | 55.0 | 18  | 45.0 |
| Total  | 33 | 42.3 | 45  | 57.7 |

Source: own elaboration.

**Table 2.** Use of medication by boys diagnosed with ASD versus boys not diagnosed with ASD

| Groups | Use of medication |  |
|--------|-------------------|------|
|        | No | %    | Yes | % |
| N      |    |      | N   | % |
| RRB    | 22 | 59.5 | 15  | 40.5 |
| ASD    | 24 | 60.0 | 16  | 40.0 |
| Total  | 46 | 59.7 | 31  | 40.3 |

Source: own elaboration.
of exploring evidence for this new clinical group, we acknowledge our study’s limitations. This is a preliminary study, and the definitions of the RRB group are still undeveloped. It is highly important to continue to conduct additional research in order to better characterize this group, and plan intervention treatments adjusted to this unique group of children who are not diagnosed with ASD but nevertheless have RRBs.

Conflict of interest

There are no conflicts of interest to declare.

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