Block design reconstruction skills: not a good candidate for an endophenotypic marker in autism research

Jonge, Maretha de; Kemner, Chantal; Naber, Fabienne; Engeland, Herman van

Veröffentlichungsversion / Published Version
Zeitschriftenartikel / journal article

Zur Verfügung gestellt in Kooperation mit / provided in cooperation with:
www.peerproject.eu

Empfohlene Zitierung / Suggested Citation:
Block design reconstruction skills: not a good candidate for an endophenotypic marker in autism research

Abstract Superior performance on block design tasks is reported in autistic individuals, although it is not consistently found in high-functioning individuals or individuals with Asperger Syndrome. It is assumed to reflect weak central coherence: an underlying cognitive deficit, which might also be part of the genetic makeup of the disorder. We assessed block design reconstruction skills in high-functioning individuals with autism spectrum disorders (ASD) from multi-incidence families and in their parents. Performance was compared to relevant matched control groups. We used a task that was assumed to be highly sensitive to subtle performance differences. We did not find individuals with ASD to be significantly faster on this task than the matched control group, not even when the difference between reconstruction time of segmented and pre-segmented designs was compared. However, we found individuals with ASD to make fewer errors during the process of reconstruction which might indicate some dexterity in mental segmentation. However, parents of individuals with ASD did not perform better on the task than control parents. Therefore, based on our data, we conclude that mental segmentation ability as measured with a block design reconstruction task is not a neurocognitive marker or endophenotype useful in genetic studies.

Key words high-functioning autism – autism – neuropsychology – psychology – cognitive style – parents

Introduction

Autism spectrum disorders (ASD) are developmental disorders that are defined on the basis of behavioral symptoms: qualitative impairments in social and communicative development, with restricted and repetitive activities and interests [4]. A large body of evidence suggests that the heritability for autism is high, but finding the genes appeared to be difficult and so far no single gene has been identified [43]. The genetic liability in autism is reflected in behavioral features sometimes found in first degree relatives that are similar but milder to those found in autism. This is referred to as the broader phenotype of autism [6, 8]. The search for genes is hindered by the fact that there is no known neurobiological marker for autism. Numerous studies, therefore, search for neurocogni-
tive features of the disorder, both in autistic individuals as well as in their first-degree relatives. Neurocognitive characteristics may be more closely linked to underlying brain anomalies and genetic factors than the behavioral phenotype [22].

One of the leading theories of neurocognitive functioning in autism suggests that autism is characterized by a weak central coherence. Weak central coherence refers to a detailed-focused information processing style in which features are perceived at the expense of the global configuration [15, 16]. As a consequence, weak central coherence predicts relatively poor performance on tasks requiring the integration of stimuli in context, but good performance on tasks that require attention to details (see [21] for a review). An example of a task that requires the suppression of the global configuration in order to process the information in a detailed fashion is the block design reconstruction subtest of the Wechsler Intelligence Scales. The construction of a block design requires mentally breaking up a pattern in separate parts and then reconstructing it by synthesizing the blocks into the original design. Autistic children and adults were found to perform relatively good on this task [9, 14, 17, 18, 20, 37, 41, 42, 44]. Their superior performance is explained by a specific asset in mentally segmenting the designs. This was shown in an elegant experiment of Shah and Frith [44], in which the reconstruction time of unsegmented and pre-segmented designs were compared within individuals. The reconstruction time difference between these pattern types was significantly smaller in individuals with autism compared to control individuals, denoting better mental segmentation skills. However, in Asperger Syndrome or high-functioning autism relative good performance on block design tasks in comparison to other tasks is not consistently found [12, 23, 32, 35, 41, 49]. Hence, the peak in performance on block design might be related to the cognitive level of individuals with autism.

There is some evidence that weak central coherence is part of the broader phenotype of autism. For example, parents of autistic individuals are found to perform better than control parents on the Embedded Figures Test, a task that requires the perception of details [7, 11, 19]. With regard to block design, superior performance on the block design subtest of the Wechsler scales relative to other subtests was not found in parents of autistic individuals [13, 40, 47]. Yet, Happé and coworkers [19] found mental segmentation superiority using the task of Shah and Frith [44] in fathers of autistic individuals compared to fathers of control families. However, in mothers or siblings of the autistic children, mental segmentation skills were not found to be significantly better than in matched control individuals [19].

Accordingly, although superior performance on block design tasks is frequently found in autistic individuals, it is not consistently found in higher functioning individuals and first degree relatives. The findings in samples including individuals with lower intellectual abilities might have been influenced by other factors such as motivational factors, uneven IQ profiles and difficulties to find a comparison group that is matched on both chronological age and IQ. In the present study, we aimed to investigate whether superior block design reconstruction skills and mental segmentation abilities are indeed phenotypic features of autism and the broader phenotype. We therefore assessed a sample of high-functioning individuals with ASD and their parents and compared them to normal control groups that were matched on sex, age and IQ. In order to maximize the chance for finding a neurocognitive reflection of a genetic liability for ASD, the individuals with ASD and their parents in the present study were all from multi-incidence families, i.e. families in which two or more siblings are diagnosed with ASD.

If weak central coherence is exhibited in superior mental segmentation skills, sensitive tasks would be needed to manifest an endowment in high-functioning individuals. We chose to use a block design task developed by Akshoomoff and Stiles [3], which has the advantage that it consists of different pattern types that vary in level of complexity and perceptual cohesiveness. We presented them in an unsegmented and a pre-segmented condition, according to the task used by Shah and Frith [44] in order to tease out mental segmentation ability. We videotaped all assessments in order to make blind rating possible and to enable the use of a computer program for accurate measurement of the reconstruction time and errors made.

We hypothesized that, compared to control individuals, high-functioning individuals with ASD and their parents, would show superior performance in reconstructing the designs especially in the most difficult condition with the highest level of perceptual cohesiveness, requiring the greatest amount of mental segmentation ability. We also expecteded the differences in performance between unsegmented and segmented patterns to be smaller in individuals with ASD and their parents than in the matched control groups. Since previous studies found behavioral and cognitive characteristics of the broader phenotype to be more prominent in males than in female relatives [5, 6, 8, 19, 38, 39, 48] we expected to find enhanced performance mainly in
the group of fathers and probably not in the group of mothers.

**Method**

**Participants**

Families with at least two children with ASD who participated previously in a genetic study of autism (by the International Molecular Genetic Study of Autism Consortium) were invited to take part in the present study. Recruitment for this previous genetic study took place in the outpatient and inpatient clinics of the Department of Child and Adolescent Psychiatry of the Academic Medical Centre in Utrecht, other centers in The Netherlands, and among members of the Dutch associations for parents of autistic individuals. Diagnostic information on the autistic individuals was obtained by means of an extensive phenotypic examination, including the Autism Diagnostic Interview Revised (ADI-R) [28, 31], Autism Diagnostic Observation Schedule (ADOS) [29, 30], The Vineland Adaptive Behavior Scales [46], and a medical assessment (screening for chromosomal abnormalities and fragile-X). Additional information was obtained from medical records. Reasons for exclusion were known medical disorders or sensory impairment.

For the present study, the first 30 families in which at least two children had a clinical diagnosis of ASD and met criteria on the ADI-R and ADOS were invited to take part and 28 families were willing to participate. Both parents and children older than 7 years of age and with an IQ of at least 70 were included. Additional psychometric data were obtained by administering a four-subtest short form of the Dutch version of the Wechsler Scales [24]. Among parents there is missing data on three fathers and two mothers who could not come to our center for assessment or where video recording of the assessment with the block design task failed. Twenty-five of the 56 children with a clinical diagnosis of ASD met the inclusion criteria (age > 7 and IQ > 70). Two additional children were excluded, due to lack of cooperation during assessment or refusing video recording. Twelve of the participating individuals had a clinical diagnosis of autism and met criteria for autism on the ADI-R and the ADOS. Eleven individuals met criteria for a spectrum disorder on the ADI-R (defined as falling one point short on one of the domains) or ADOS. Their classifications on the basis of the DSM-IV were Autism (n = 1), Asperger Syndrome (n = 7) and PDD-NOS (n = 3).

As a comparison group for the individuals with ASD, typically developing children and adolescents were recruited from regular schools. They were screened by telephone for behavioral or psychiatric problems, or ASD in their family, and were then assessed with the Child Behavior Checklist (CBCL) and the Teacher Report Form (TRF) [1, 2, 50–52]. Individuals with psychiatric or behavioral problems or with first or second-degree family members with ASD were excluded (n = 3). Thirty-seven children participated in the study, but the data of one child was lost because the assessment was not visible on video. The clinical and the control group were matched for gender, mean age, and IQ.

As a comparison group for the parents of the children with ASD, parents of a child with down syndrome (DS) were recruited from the community through the Dutch Down syndrome Parent Association. The reason for choosing parents of children with another disability as a comparison group was because of research questions that were addressed in a separated study and not reported on in this paper. The parent groups were also matched for gender, mean age, and IQ. There were 30 control parent couples participating in the study, but missing data on three control parents: one father could not come to our center for assessment and videotaped data were lost for two mothers. Altogether, 167 individuals participated in our experiment.

All participants had normal or corrected to normal vision. The experimental protocol was approved by the Medical Ethics Committee of the University Medical Centre Utrecht and informed consent was obtained before testing from all parents and from children older than 12 years.

Table 1 shows the mean age of all individuals and Table 2 shows the mean IQ scores on the short form of the Wechsler scales for the subject groups. There were no significant differences in age, TIQ, VIQ or PIQ between any of the groups and their control groups. Also, no differences were found between the fathers and the mothers of the ASD and control groups.

**Materials**

Patterns from the task developed by Akshoomoff and Stiles [3] were used. Materials consist of the set of red

<table>
<thead>
<tr>
<th>Table 1 Subject characteristics: mean age (and SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>ASD group</strong></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>N</td>
</tr>
<tr>
<td>Mean</td>
</tr>
<tr>
<td>Children/adolescents</td>
</tr>
<tr>
<td>Parents</td>
</tr>
<tr>
<td>Fathers</td>
</tr>
<tr>
<td>Mothers</td>
</tr>
</tbody>
</table>
and white blocks of the Wechsler Scales. Each block has two red sides, two white sides and two sides that are half red and half white with the division oriented along the diagonal. The target-patterns were printed in red and white and collected in a binder. The first set of six patterns require four blocks \((2 \times 2\) squares) to construct the pattern, the second set of 6 patterns require nine blocks \((3 \times 3\) squares). There were two different pattern types: local and global patterns. For local patterns, perceptual cohesiveness (the number of block edges bordering on a neighboring block edged with the same color) was low, while in the global designs there was a maximum degree of perceptual cohesiveness. Within each set there were three local and three global patterns, which were presented in alternated order. Then a third and fourth set of pre-segmented patterns were presented. These patterns were exactly the same as in the first and second set and presented in the same order, but the block-edges were outlined with a black line. There were 24 patterns altogether plus a 4-blocks and a 9-blocks practice item.

### Procedure

All participants were individually tested in a quiet room. First, the participants were shown four blocks and it was explained that all blocks were colored the same. The different sides of the blocks were shown and the practice item was presented. All participants were well able to construct the practice item. Then the task conditions were explained to the subject and they were encouraged to reconstruct the patterns as quickly as possible. The administration of the task was videotaped for later data analysis. Although the videos were used to measure construction time, a stopwatch was used during the task, to provide extra non-verbal pressure on the participants to reconstruct the patterns as quickly as they could. Participants were allowed maximally 90 s to construct the 4-block patterns and 180 s for the 9-block patterns. After six items the additional blocks were added, so that the subject had nine in total and a practice item with nine blocks was presented. Again, all participants were well able to construct this design.

### Scoring

Before each test, a subject identification number was shown in front of the camera, which made it possible to code the videos blind to group status. Videotapes were coded by means of the “The Observer” [34]. This is a software system for recoding, coding and analyzing frequencies and durations of observed events. The use of this video tape analyses system enables coding behavior at different VCR playback speeds, while maintaining a proper time reference. This enables an exact coding (in hundredths of seconds) of the start and end of each occurrence of behavior.

For each design, the final result (whether the design was correctly reconstructed within the given time) and the time taken to reconstruct the pattern, was recorded. In addition, the number and type of errors made during reconstruction were recorded. Most of these errors were self-corrected within time limits. Three types of errors made during reconstruction were defined. Color errors refer to placing a block with the wrong colored side up (for instance red-side instead of white-side). Matrix errors refer to breaking the \(2 \times 2\) or \(3 \times 3\) matrix by placing for instance four blocks on a row. Block rotation errors refer to red-white block placed with the diagonal oriented in the incorrect direction. The block design task was coded by two observers, who were blind to the diagnostic status of the participants. To test the inter-rater agreement of the coding, 33 videotapes (=20%) were coded twice by the independent coders. The agreement corrected for chance was 0.72 (Cohen’s Kappa) which is usually interpreted as substantial agreement [27].

### Data analysis

The statistical package SPSS (version 11.5 for Windows) was used to analyze the data. The accuracy
score was defined as the total number of patterns correctly reconstructed. The participants were well able to reconstruct the patterns correctly. Since the accuracy data were not normally distributed a 2-tailed Mann-Whitney U was used to compare the mean number of correctly constructed designs between individuals from ASD families with individuals from the matched control group. The mean reconstruction time was defined as the mean time needed to reconstruct the patterns and was calculated for the different pattern types separately. Mean number of errors and error types made during reconstruction of the designs were calculated for the different pattern types separately. Mean number of errors and error types made during reconstruction revealed a significant interaction effect for Number × Group F(1, 57) = 6.51, P = 0.049. Partial interactions showed a Group × Pattern Type effect in the unsegmented 9-block patterns F(1, 57) = 5.12, P = 0.03. However, when tested out, it was found that both groups reconstructed the local patterns in this condition significantly faster than the global patterns, although the difference was smaller in the ASD group (individuals with ASD: mean difference local-global = 12.2 s, t = 4.66; matched control group: mean difference local-global = 20.0 s, t = 6.51).

The planned comparisons revealed that there was no significant difference between the groups on the most difficult task condition (the 9-blocks, global, unsegmented patterns). The difference score (9-blocks, global patterns in the unsegmented condition versus the segmented condition) was compared between groups. The difference in reconstruction time between these conditions did not reach significance when we compared the ASD group to the matched control group.

## Reconstruction time in the children groups

When comparing the group of individuals with ASD with the matched control group, analysis of variance revealed an interaction effect on the borderline of significance between Group × Number × Segmentation × Pattern Type F(1, 57) = 4.03, P = 0.049. Partial interactions showed a Group × Pattern Type effect in the unsegmented 9-block patterns F(1, 57) = 5.12, P = 0.03. However, when tested out, it was found that both groups reconstructed the local patterns in this condition significantly faster than the global patterns, although the difference was smaller in the ASD group (individuals with ASD: mean difference local-global = 12.2 s, t = 4.66; matched control group: mean difference local-global = 20.0 s, t = 6.51).

The planned comparisons revealed that there was no significant difference between the groups on the most difficult condition requiring the highest level of mental segmentation ability (i.e., 9-blocks, global, unsegmented patterns). Second, a difference score was calculated, based on the methods used in the study of Happé, Briskman and Frith [19]. The difference score referred to the difference in reconstruction time between unsegmented en pre-segmented designs in the 9-block, global condition. All significance tests presented are two-tailed. Statistical significance was defined as \( P < 0.05 \).

## Results

The accuracy level, that is, the percentage of patterns constructed correctly, was high for all individuals. The mean number of correctly reconstructed designs was 23.8 (SD = 0.4) in the ASD group and 23.4 (SD = 0.8) in the control group. In parents the mean number of correctly constructed designs was 23.5 (SD = 0.8) in the ASD-parent group and 23.3 (SD = 1.5) in the matched control group of parents. There was no significant difference between individuals with ASD and individuals from the control group in the number of correctly constructed designs. Also, no significant differences were found between the fathers or mothers of the individuals with ASD and the fathers or mothers of the individuals with DS.

### Reconstruction time in parents

For the groups of parents no significant interaction effect was found between any of the manipulations and the between-subjects factor Group (ASD parents vs. Control parents). In fact, the reconstruction times were very similar in both groups in all conditions as is shown in Fig. 1. For fathers, repeated measures also revealed no significant interaction effect between any of the manipulations and the between-subjects factor Group (ASD fathers vs. Control fathers). Planned comparisons also did not show differences.

### Errors made during construction

An analysis of variance on total number of errors made during reconstruction revealed a significant interaction effect for Number × Group F(1,
Further analysis showed that there was a significant Group interaction effect for the 9-block designs only $F(1, 57) = 13.1, P < 0.01$, indicating that the individuals in the ASD group made significantly fewer errors (mean number of errors = 1.86; SD = 3.4) than the individuals in the control group (mean number of errors = 5.57; SD = 4.9). In parents however, there were no group effects (mean number of errors 9-block condition parents ASD group = 4.1; SD = 4.5; parent control group = 3.6; SD = 5.5).

Further analysis showed that there were specific kinds of errors made more often by individuals in the control group than by individuals with ASD. With respect to the number of Color errors we found no significant differences between the ASD-group (mean number of errors = 0.77; SD = 2.4) and the matched control group (mean number of errors = 1.43; SD = 2.1), nor between the parent groups. With respect to the number of Matrix errors, a significant interaction effect found was for Number $\times$ Group $F(1, 57) = 6.51, P < 0.013$, but further analysis for the 4- and 9-block conditions showed no significant groups effect (mean number of errors 9-block condition ASD group = 0.45; SD = 1.1; control group = 1.11; SD = 1.8). No significant differences were found between the parent groups.

In addition, for the number of Block rotation errors made during reconstruction, a significant interaction effect was found for Number $\times$ Group $F(1, 57) = 8.52, P < 0.01$. Further analysis showed that there was no interaction effect for the 4-block patterns between groups, but a significant group effect for the 9-block patterns $F(1, 57) = 15.84, P < 0.01$, indicating that the individuals with ASD made significantly fewer Block rotation errors (mean number of errors = 0.64; SD = 1.3) than the individuals in the control group (mean number of errors = 3.03; SD = 3.3). In parents, there were no significant differences between groups (mean number of Block rotation errors 9-block condition parents ASD group = 1.9; SD = 2.4; parent control group = 1.6; SD = 1.9).

**Discussion**

This study investigated whether superior performance on a Block Design reconstruction task, could be identified in high-functioning individuals with ASD from multi-incidence families compared to a matched control group. In addition we investigated whether superior performance was found in the parents of the individuals with ASD compared to a matched control group of parents. Since our sample consisted of high-functioning individuals with ASD and their parents, in whom these cognitive assets might be subtle, compared to matched control groups, we used a task that was thought to measure segmentation skills in the most sensitive way.

Contrary to our expectations, we found no significant differences in reconstruction time between high-functioning individuals with ASD and matched control individuals. Absolute measures (planned comparison between reconstruction time on the most challenging condition of the task: 9-block unsegmented global patterns) did not reveal significant differences in performance between groups. We used two relative measures. Reconstruction time differences were compared between the local and global patterns in the unsegmented 9-block condition. Both groups however, were significantly faster in reconstructing the local than the global patterns, although the difference was somewhat smaller in the ASD group. In addition we did a planned comparison between reconstruction time of the 9-block global unsegmented and the segmented patterns. We did not find the difference in reconstruction time between these conditions to be significantly smaller in the ASD group than in the matched control group. Hence, there were no indications that the individuals with ASD were less aided by the pre-segmentation of the designs in comparison to the control group.

We expected that superior performance on the Block Design task would not only be reflected in the velocity in reconstructing a pattern but also in the accuracy with which the individuals position a block during the process of reconstruction. The assumption was that individuals with superior mental segmenta-
tion abilities place a block in the correct position within a design right away. While contrast, individuals with fewer mental segmentation skills, would more frequently place a block in an incorrect position initially and adjust that subsequently during the process of reconstruction. Indeed, we found that individuals with ASD made significantly fewer errors during the reconstruction of the 9-block patterns than control individuals. The differences between individuals with ASD and individuals from the matched control group were most striking in a specific type of error: block rotation errors. This error type refers to a diagonal block face rotated in an incorrect direction. In contrast, no differences were found between groups in color errors e.g. placing a red-sided block while a white-sided block was required or in matrix groups in color errors e.g. placing a red-sided block.

Individuals with ASD have significantly less incorrect shapes before finding the right one, compared to matched controls [11]. These findings are consistent with a study in which the Embedded Figures Test was used in a high-functioning sample of individuals with ASD and their parents. When looking at the number of errors during the process of searching for the correct shape, both individuals with ASD and their fathers pointed out significantly less incorrect shapes before finding the right one, compared to matched controls [11].

In the present study, we also looked at the performance of parents on the Block Design task, but did not find any evidence for superior performance in parents. Parents from ASD families were not faster than the control group of parents in reconstructing the designs, not even in the most strenuous pattern type condition. When looking at difference scores between local and global, or unsegmented and segmented patterns, in order to focus specifically on mental segmentation ability, parents of ASD families, did not stand out either. In addition no differences in the number or specific type of errors were found between the parent groups. It is previously found that behavioral characteristics of the broader phenotype are more common in males than in female relatives [5, 6, 8, 38, 39, 48] and since there is some evidence that this also extends to cognitive features [19], we looked at the performance of fathers separately. However, there were no differences between fathers from children with ASD and control fathers on any of the measures.

Taken together, we conclude that in our sample of high-functioning autistic individuals, we could not replicate weak central coherence as reflected in faster block design reconstruction performance in comparison to a matched control group. Neither absolute, nor relative measures revealed significant differences in reconstruction time between the two groups. This is in line with previous studies using different versions of Block Design tasks in high-functioning individuals with autism or Asperger's syndrome. In some of these studies a peak on the Block Design task compared to other tasks was relatively small [17, 20, 45], while in other studies it was not found at all [12, 35, 36, 41, 49]. Although there are also studies that reported significant higher scores on the Block Design task in high-functioning subjects compared to controls [10, 33] most studies found significant better performance in lower functioning individuals with autism but not in high-functioning individuals [32, 41, 44]. In the present study the number of times a block was placed in the correct location and orientation at first attempt appeared to be a more sensitive measure of weak central coherence or mental segmentation ability. Previous research showed that a larger number of errors made during the reconstructing block designs are associated with weaker visuospatial skills in typically developing children and adults [25, 26].

We found no evidence for a reflection of weak central coherence in superior mental segmentation skills in parents of autistic individuals, nor in fathers of autistic individuals separately. This is not in line with the study of Happe et al. [19], who found fathers of autistic individuals to be significantly faster on unsegmented patterns, but not on pre-segmented patterns, compared to fathers of typically developing children. Our results replicate however, the results of two studies not finding fathers nor mothers of autistic individuals to stand out on the Block Design task [13, 47].

The main limitation of our study is that our sample includes a relatively modest number of individuals, due to the fact that we recruited multi-incidence families. The lack of differences between groups might be a result of a lack of power. However, compared to studies that found superior block design performance in their sample, the size of our samples was not smaller than most of these studies. In addition, the reconstruction times of the parent groups are very much alike and do not suggest possible differences in performance that might become significant when adding a reasonable number of individuals to our sample.

A particular strength of this study lies in the measure used to assess block design reconstruction performance. We choose a task that made fine-tuned measuring of subtle differences in performance possible. Low perceptual cohesive (local) patterns and high perceptual cohesive (global) patterns, could be
presented in alternated order without revealing prompts for a strategy to mentally segment the patterns. We did not only measure reconstruction time but the number and type of errors made during the process of reconstruction as well. Another strength of the present study is that we choose to use a software system designed to code observational data, in order to be able to score the data blind to group status. Therefore, all measurements were recorded on videotape. This method also made very accurate time measurements possible.

In conclusion, there are some indications for good mental segmentation abilities, as measured with a block design reconstruction task, shown by a lower number of errors made during reconstruction, in the high-functioning individuals with ASD compared with a matched control group. However, superior mental segmentation ability was not evident when looking at reconstruction time of the most difficult patterns or the extent to which subjects were aided by pre-segmentation. Furthermore, dexterous mental segmentation ability did not extend to the broader phenotype. Therefore, based on our data, we might conclude that it is not a neurocognitive marker or endophenotype useful in genetic studies.

References


