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Kinetic form discrimination in Prader-Willi syndrome

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Abstract

Discrimination of the shape of motion-produced forms generated by random elements (i.e. second-order stimuli varying in element density and temporal correlation) was tested in four groups: (1) subjects with Prader-Willi syndrome (PWS), chromosome 15q deletion subtype; (2) subjects with PWS, uniparental maternal disomy (UPD) subtype; (3) equivalent non-PWS controls; and (4) normal controls. The performance of the normal controls exceeded that of all other groups (78% correct, $P < 0.009$). The PWS deletion (66%) and the equivalent control groups (59%) did not differ($P < 0.95$). The U PD group performed significantly less well (38%, $P < 0.04$) than all the other groups. The performance of the PWS deletion and equivalent control groups is consistent with other data indicating that these populations encounter difficulty meeting the processing demands posed by second-order stimuli. The inferior performance of the UPD group may be attributed to receiving two active alleles of a maternally expressed gene influencing neural development. One candidate is the ubiquitin protein ligase gene (UBE3A), which is maternally expressed only and localized to the 15q region. Other possibilities include the requirement of a paternally expressed gene, residual mosaic trisomy 15 in the brain tissue or complex interactions including specific ratios of differentially spliced gene products.

Keywords

global motion; intellectual disability; kinetic form discrimination; Prader-Willi syndrome; second-order stimuli; visual perception

Introduction

Among those syndromes induced by genetic anomalies associated with intellectual disability (ID), Prader-Willi syndrome (PWS) is notable because of both the mechanism of genetic transmission through imprinting and the diversity of its effects. Occurring with a frequency

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of one in 10 000–20 000, PWS involves genetic defects and imprinting on the chromosome 15q11-q13 regions, and is the most common syndromal cause of marked obesity in human adults. During infancy, clinical signs include hypotonia, hypogonadism, feeding difficulties and failure to thrive. In early childhood, hyperphasia and obesity develop concurrently with mild to moderate ID. The majority of afflicted individuals have a deletion in the 15q11-q13 region that is derived paternally. The others usually show uniparental disomy (UPD), i.e. both chromo-some 15s are from the mother (Nicholls *et al.* 1989; Butler 1990; Cassidy *et al.* 1997).

The present study investigated visual perception in people with PWS, a topic that has received relatively little research attention. The specific perceptual capacity examined involved discrimination of forms generated by arrays of random elements engaged in apparent motion. Because this class of stimulus may not be familiar to all readers, some comments about specific attributes may be helpful. The standard stimulus for vision and visual perception can be defined in terms of differences in the intensity of light impinging on specific areas of the retina which produce percepts of edges and lines. However, similar percepts can be induced by the coordinated actions of groups of small, random elements changing along a continuum of either time or space. Detection of the global change in sets of these elements is the critical variable for the induction of percepts rather than the specific physical characteristics of the individual elements. An interest in these kinds of perceptual phenomena is not new. The core assumption of Gestalt psychology focuses on such effects, as exemplified by the so-called laws of perceptual organization, such as perceptual closure, completion and common fate. The advent of computer technology made it possible to generate and rapidly manipulate the parameters of large sets of stimulus elements which induce global percepts. Two notable products of this line of inquiry, which developed independently of classical Gestalt theory, are the random-element stereogram (RES) and the random-element kinematogram (REK; Julesz 1960, 1971). The RES consists of two arrays or matrices of random elements, each of which is presented to a separate eye. The view of elements seen by one eye contains no forms or contours, but when the pair of arrays is viewed stereoscopically by an observer who possesses stereopsis, the visual system can detect small differences in the spatial position of a subset of elements in the two arrays. Detection of these differences in the microstructure of the arrays induces the percept of a stereoscopic form with distinct edges and a seemingly palpable surface located at a specific position in depth. The form can be seen only by the observer who possesses stereopsis. It is generated by the binocular visual system and exists phenomenally without a direct physical counterpart. The form is a product of computations based on the information inherent in the variations in the structure of the random element arrays. For this reason, such forms are often referred to as global, hence the terms global stereopsis and motion.

Global forms are also induced by REKs by processing analogous to that operative for stereograms. The starting point for a REK is an array of random elements, each of which is displaced in a random direction by a small amount over time. The arrays are presented sequentially in discrete frames using video or film image generation methods. The spatial displacement of each element over successive frames produces apparent motion of the elements that generates a random pattern similar to the noise seen on an untuned video channel. However, if a subset of the elements in the random array is made to move in the

same direction across frames, the visual system detects this correlated movement embedded in the array of random elements, and the observer perceives a form with distinct edges and surfaces similar to the global form produced by stereograms. The form induced by the kinematogram is produced by computations performed on the random elements and does not exist physically. It can be seen only as long as the apparent motion in the array is present.

Considerable interest in random element stimuli, and particularly, RES and REK, has been generated ever since their inception, and continues to be a major topic in basic research on vision. Discussions of these phenomena are now a standard component of introductory texts on perception (e.g. Sekuler & Blake 1994).

For the purposes of the present study, three of their well-established characteristics should be noted. First, at the level of behavioural or psychophysical analysis, it is clear that global stimuli are functionally quite similar to their physical counterparts defined by differences in luminance, i.e. they are seen quickly without engaging attention or other cognitive variables, and generate many classic perceptual phenomena such as masking and aftereffect induction. Indeed, in at least two ways, they are more robust than physical stimuli. When element density is reduced in RES and REK displays, the surfaces of global forms and their edges remain visible because a perceptual completion or filling-in process occurs that functionally replaces missing elements and maintains the integrity of the form. They can still be seen even when almost all of the initial elements (e.g. 97%) have been removed. Furthermore, the forms are relatively immune to optical degradation that would blur physically defined edges and render such stimuli difficult to perceive. Presumably, the neural origin of the global forms renders them immune to blurring of the retinal image.

Secondly, at the neurophysiological level, it is clear that random element stimuli are processed in the cortical areas devoted to an early stage of visual processing (e.g. V_1, V_2 and MT). These data are consistent with the psychophysical evidence indicating that these kinds of perceptual phenomena are processed automatically and can conceptually be regarded as being part of the domain regarded as cognitively impenetrable (Pylyshyn 1999).

Finally, in terms of formal models of visual processing, random element stimuli pose problems in the sense that the generation of global forms requires additional computational processes not necessary for the processing of forms defined by differences in luminance. One possibility under active consideration is that there are two independent parallel processing modes for each of the two stimulus classes. In recognition of such differences, it has been suggested that random-element stimuli in general should be designated as second-order stimuli (Cavanagh & Mather 1990).

The potential burden on computation resources posed by random-element stimuli is one underlying reason for examining the ability of special populations to process these kinds of stimuli effectively. Prior research has revealed impairments in processing when density and correlation are reduced (Fox & Oross 1988, 1990, 1992). An extension of the inquiry to individuals with PW S constitutes a natural next step.

Subjects and methods

Subjects

The critical aspect of the present investigation involved the recruitment of people with PWS in sufficient numbers to yield statistically meaningful sample sizes. These subjects were obtained through a research programme devoted to an assessment of PWS in which individuals were recruited, nationally, and invited to visit Vanderbilt University, Nashville, Tennessee, USA, for 2–3 days of comprehensive testing, including genetic classification. The present study was reviewed and approved by the Institutional Review Board of Vanderbilt University. Written consent was obtained from all relevant individuals.

The technique used for classification included high-resolution chromosomal analysis, *in situ* hybridization, microsatellite DNA analysis with polymerase chain reaction (PCR) and methylation PCR procedures, as described previously (Butler *et al.* 1986; Mutirangura *et al.* 1993; Butler *et al.* 1996; Spritz *et al.* 1997; Kubota *et al.* 1997; Muralidhar & Butler 1998). These analyses insured the unequivocal assignment of individuals into the UPD ($n = 19$) and deletion ($n = 24$) subgroups. At the same time as the PWS group was assembled, participants willing to serve in an equivalent control group ($n = 22$) were recruited. The purpose of this group was to provide data from people without PWS who had undergone the same testing protocols as the subjects with PWS, and were equivalent on the variables of intelligence, age and fat mass. In Table 1, the degree of matching among groups on the above variables is given.

To provide baseline data for evaluating the performance of the PWS and equivalent control groups, a second control group was assembled composed of individuals with normal intelligence($n = 14$) and presumably free of relevant clinical syndromes. These were recruited from the graduate and undergraduate student population at Vanderbilt University. The majority of normal controls were not experienced observers in experiments on perception and received no explicit incentive for performing well.

The groups were not equated on peripheral visual capacities such as visual acuity, although subjects were free to wear their corrective lenses. Visual capacity was not equated because it is not a relevant variable for at least two reasons. First, the percepts induced by second-order stimuli are quite resistant to optical blur, presumably because they are generated by cortical mechanisms which extract information from the arrays of random elements. Secondly, the stimulus parameters which influence discrimination performance involve processing the organization or global character of individual elements, such as their correlation. The critical variable is the overall spatial position of the elements, collectively perceived, rather than the optical clarity of single elements.

Random-element stimuli

Some general comments about the generation and appearance of random-element kinematograms may be useful before describing the specific display used in this inquiry. When kinematograms are generated by computers, arrays of small elements, i.e. dots, fill the display surface of a CRT monitor operating in the raster scan mode. The surface can be regarded as being divided into a matrix of cells, each of which may contain an element that

fills the cell. When half of the cells are filled, which is the standard or typical condition, the density of elements is designated as 50%. When two or more matrices or arrays of elements are successively presented, as in the case of successive frames in raster displays, differences in the x- and y-positions of individual elements between frames induce apparent motion. If the position of individual elements varies randomly between frames, i.e. each element assumes different left, right, up and down positions, an overall perception of apparent motion of the display is seen resembling the visual noise visible on an untuned video channel or the Brownian motion of molecules. This kind of motion is often called incoherent or uncorrelated. If a subset of the elements is instructed in effect by the computer program to shift in a common direction, such as all moving to the left, a percept of a kinetic form is seen. The shifts in correlative apparent motion are said to be coherent or correlated. The detection in processing in such shifts by the visual system is responsible for the emergence of the form. If the apparent motion stops, then the form disappears. When all of the elements in the x- and y-regions of the display defining the form move in the same direction, the coherence or correlation is designated as 100%. Reductions in correlation are defined by reducing the proportion of elements in the area that is shifting in a common direction, with the remainder of the elements shifting in random directions. Element density and correlation are two stimulus parameters identified early on (e.g. Julesz 1971) which influence the discriminability of kinetic forms.

The specific kinematogram stimuli used in the present investigation were generated by a DOS-based computer program developed by B. Bertenthal and described previously (e.g. Fox & Oross 1990). They were displayed on an achromatic monitor, which at a viewing distance of 90 cm sub-tended 12 × 17.2°. The duration of each frame or raster was approximately 17 ms, and the duration between successive frames was 34 ms. The global form generated, for which discrimination was required, was configured as the letter ‘E’. It consisted of four rectangles joined to form the letter and appeared in the centre of the display for an unlimited duration with a continual recycle duration of eight frames. The form could appear on any given trial in any one of four equally probable orientations (i.e. up, right, down or left), thereby permitting forced-choice discrimination of orientation to be used as the indicator response.

Design and procedure

The goal of this experiment was to determine the discriminability of the E-form as a function of reductions in element density and temporal correlation. The computer program for generating the kinematograms could present forms varying in six levels of density (i.e. 50%, 25%, 12%, 6%, 3% and 1%), and with seven correlation values (i.e. 100%, 85%, 70%, 55%, 40%, 25% and 10%), thereby yielding, in factorial combination, a set of 42 forms. The discriminability of the complete set has been assessed in prior research (Fox & Oross 1992). The above authors revealed that reductions in both variables impaired discrimination, with correlation having a significantly greater impact. However, the full set of 42 forms could not be used in the present experiment because of constraints on time. Instead, subsets of forms were selected to examine the effects of density reduction alone, correlation reduction alone, and for some combinations of density and correlation reduction. Three successive series were used. The first consisted of six density values (i.e. 50, 25, 12, 6, 3 and 1). The second

consisted of seven correlation values (i.e. 100, 85, 70, 55, 45, 25 and 10). The third consisted of six density/correlation combinations (i.e. 50/100, 25/85, 12/70, 6/55, 3/40 and 1/25). A shift from one series to another was made if a subject made two consecutive errors or if the subject completed the series without error.

To indicate their discrimination response, the subjects pointed to a panel containing drawings of each of the four possible orientations of the form displayed on the four quadrants of the panel. Verbal reports of orientation were also acceptable.

Two preliminary tasks were used to teach the subjects the task requirements and to determine their comprehension of the task. In one, E-forms were presented as luminance-defined forms on a video monitor and the orientation was shifted randomly over six presentations. The subjects had to indicate their perceived orientation of the form. The second task involved discriminating the direction of motion, left or right, of an array of moving vertical contours generated as a kinematogram over six presentations. All subjects made both kinds of discriminations without error and gave every indication that they comprehended the task requirements.

Results

The proportion of correct discriminations for each of the four groups is shown in Fig. 1. The significance of the differences among groups, as assessed by the Mann-W hitney *U*-test, was as follows. The performance of the normal control group was superior to that of all other groups ($P < 0.009$). Differences between the equivalent controls and the PWS deletion groups were not significant ($P < 0.95$). The performance of the PWS-UPD group was significantly worse than that of all other groups ($P < 0.004$). Since the subjects with PWS-UPD exhibited the poorest performance on the kinetic form discrimination task and had the lowest performance IQ, an analysis of covariance was employed to control for the potential effect of performance IQ on kinetic form discrimination in the three groups with reduced cognitive function. This analysis demonstrated that group differences persisted after controlling for performance IQ in these subjects ($P = 0.002$). The least-squares adjusted means for the three groups on proportion correct were 0.54, 0.65 and 0.41, respectively, for the equivalent controls and the subjects with deletion and UPD.

In Table 2, the performance of the groups is given as a function of the three stimulus series, density reduction alone, correlation reduction alone, and combinations of density and correlation reduction. The pattern of results supports the hypothesis that discriminability of the E-form is increasingly impaired as a function of reductions in correlation and density. This is also supported by an analysis of overall errors made on specific forms. An analysis of errors made on individual forms, analogous to an item analysis used on paper-and-pencil tests, revealed that very few errors were made on the standard form composed of 50% density and 100% correlation values. At the other end of the continuum, relatively low values of density and correlation generated many errors, even among the members of the normal control group.

Discussion

The discussion of the results is divisible into two parts: (1) the inferior performance of the PWS and equivalent control groups relative to the normal control group; and (2) the inferior performance of the U P D group relative to the deletion group.

With respect to the first part, the impaired performance is similar to that observed in prior research. In their experiment 1, Fox & Oross (1988) found that adults with mild ID encountered difficulty in discriminating the shapes of global stereoscopic forms. This task does not involve the capacity for detecting stereopsis because stereopsis is required in order to see the forms. Rather, the impairment involves the spatial resolution required to distinguish one form from another (e.g. a rectangle and a square). An analysis of errors in discrimination suggested that the observers with ID perceived the forms more as blobs than shapes with well-defined edges. In contrast, the non-ID control subjects discriminated correctly among all forms without error.

The existence of separate stages, one for detection of the retinal disparity requisite for stereopsis and one for resolving the global form, is a well-known feature of global stereopsis. Nevertheless, in most instances, both stages seem to operate together, and one perceives the form in depth and can also describe its shape within about the same time interval. Given this context, the difficulty in stereoscopic form discrimination encountered by the subjects with ID was unexpected. It contributed to the idea that the requirements for neural computation posed by second-order stimuli may exceed the computational resources of these individuals.

To provide a theoretical framework for this view, the general computational model developed by Marr (1982) was invoked. In this approach, an initial processing stage called the primal sketch is dedicated to detecting the correlations among elements in space or time which are intrinsic to second-order stimuli such as global stereograms and kinematograms. A second stage called the 2–1/2D sketch computes the configuration and surface of the emergent form in x- and y-coordinates. One implication of this view is that the deficit in form discrimination involves the 2–1/2D stage and would become manifest for all classes of second-order stimuli rather than being restricted to global stereopsis. To test this implication, the discriminability of motion-defined forms induced by kinematograms was investigated (Fox & Oross 1990, 1992). These studies found substantial deficits in the discrimination of forms, a result that supports the idea of an impairment in spatial resolution resident in the 2–1/2D stage.

As a practical matter, the existence of deficits in the motion domain side-steps the methodological problems posed by the requirement that adequate binocular stimulation must be present as a precondition for inducing stereoscopic phenomena. This means that global motion phenomena can be used as an effective technique for testing second-order stimulus perception in special populations. This consideration motivated the present study of the ability of persons with PWS to discriminate forms generated by kinematograms. The results indicate that the performance of the PWS deletion subgroup and the equivalent control group are quite similar, with both groups being inferior to the normal control group. This is the

same pattern of results that has been found in previous studies. It suggests that the interpretation of a deficit in spatial resolution present at the 2–1/2D stage can be extended to the PWS population.

The related idea of an impairment or limit on neural computation is also applicable. Although that concept has been outlined only in a general way (e.g. Fox 1998), it is intended to capture an important feature of the deficit in the processing of second-order stimuli. This is the marked effect on discriminability when density and correlation are reduced in kinematograms. These reductions constitute removal of physical stimulation that supports the percept. In normal observers, some compensation for reduction occurs at the neural level that acts to maintain the configuration of the percept. The clearest example of this is the formation of subjective contours which observers report when element density is reduced. Note that no members of the normal control group made errors in the condition where only density was reduced. Presumably, subjective contours emerged that maintained the integrity of the E-form sufficiently to enable its orientation to be discriminated. The process that compensates for reduction in physical stimulation is thought to be a product of neural computation. Its absence would account for the character of the deficits which have been found. This interpretation assumes that the deficit involves an early automatic part of the perceptual system, and cannot be explained by alternative interpretations based on higher level stages related to attention, motivation or comprehension.

These kinds of interpretations have been examined in previous studies and rejected. The same arguments apply to the present study. In this regard, it is worth pointing out salient aspects of this experiment which argue against alternative interpretations. These include the use of a forced-choice response method that minimizes variations in response criteria and the screening of subjects for task comprehension. Moreover, performance was quite sensitive to variations in stimulus parameters. For some combinations of density and correlation, most subjects made correct discriminations, yet failed on other combinations, i.e. stimulus interaction occurred. However, task requirements remained the same across all stimulus combinations.

With respect to the second component of the results, the difference between the two PWS genetic subtypes, the general interpretation discussed above seems applicable. The key difference is the marked inferiority in discrimination found for the UPD group. It should be noted that an indication of such a difference between subtypes was found in a task involving global stereoscopic form discrimination. This observation was made incidental to an examination of visual capacity in PWS (Fox *et al.* 1999) and was not pursued parametrically. It partly prompted the present study wherein the use of motion avoids interpretative problems related to the capacity for stereopsis.

An interpretation of the difference between genetic subtypes arises naturally from the differences in genetic transmission between them. In the deletion case, the mother contributes one active genetic factor, while in the UPD case, the mother’s contribution would be doubled. If the factor produced some deleterious effect on neural development, one straightforward interpretation would be a greater effect for the UPD subtype. One candidate is the ubiquitin protein ligase (UBE3A), which is not normally expressed in the paternal

15q11-q13 chromosome region and is implicated in Angelman syndrome (Sutcliffe *et al.* 1997a, b). It may influence synaptic density during neural development and is normally expressed only on chromosome 15 from the mother. The presence of two active maternal copies of the gene, as in the case of the PWS-UPD subtype, could have negative consequences for the complex behavioural function of these individuals. Other possibilities include the requirement of a paternally expressed gene, residual mosaic trisomy 15 (two chromosome 15s from the mother and one chromosome 15 from the father in the brain tissue) or complex interactions including specific ratios of differentially spliced gene products.

Two related, albeit speculative ideas merit mention here. One is the various lines of evidence assembled by Eysenck (1998) which suggest links between specific components of neural development (e.g. synaptic density) and complex behavioural function. Secondly, the substrate for processing second-order stimuli is thought to be activity in a network of closely coupled neurons that maintains perception in the absence of concurrent physical simulation.

Investigation of these implications is facilitated by the considerable literature on motion perception induced by kinematograms. It includes data from human adults, children and infants, and animals in sufficient quantity to formulate a standard measure of second-order stimulus processing equally applicable across several levels of behavioural complexity.

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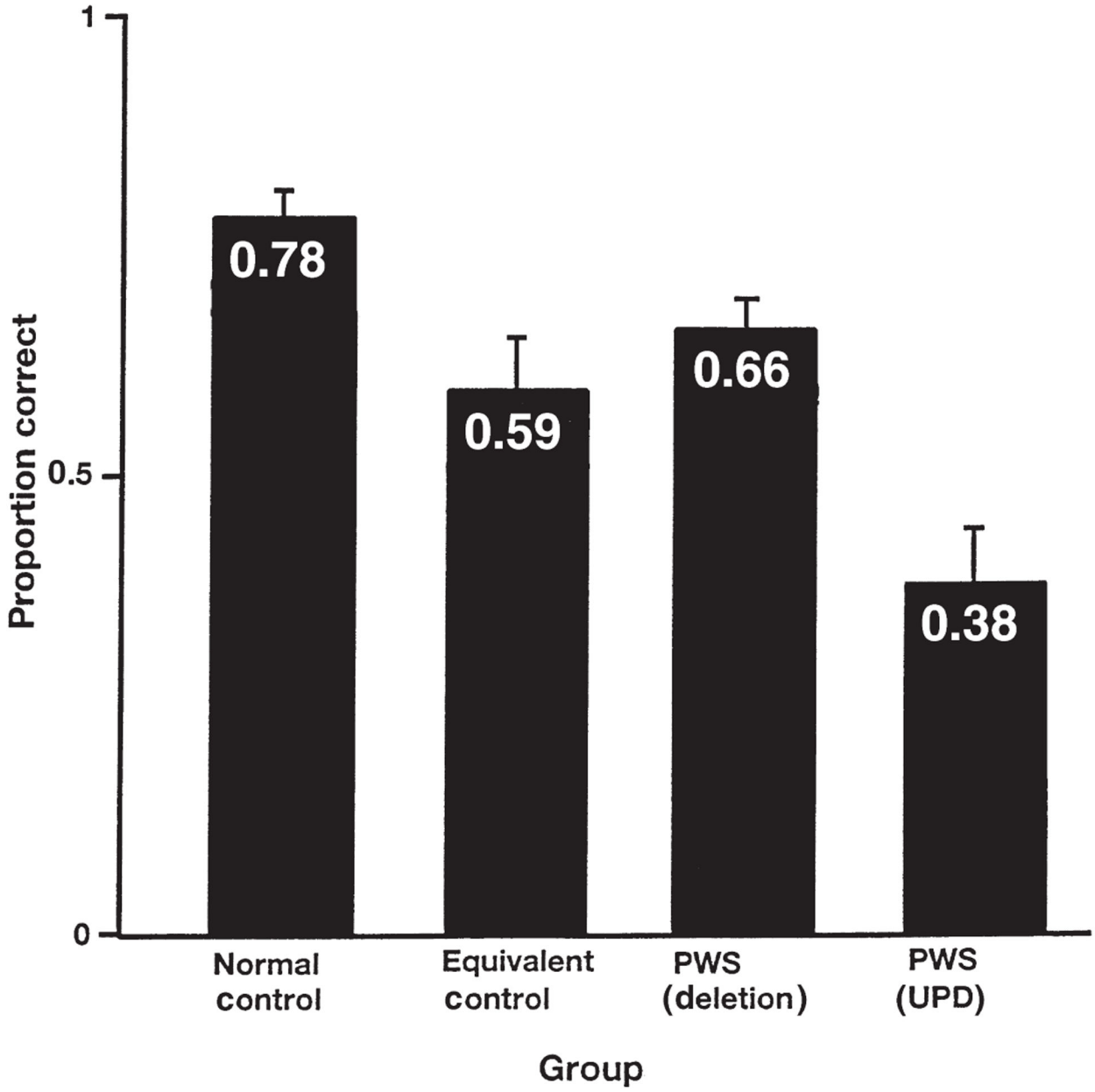

Figure 1.
Overall proportion of correct identifications for each of the four groups. The lines above the bars indicate one standard error.

Table 1

Mean number of matching characteristics (± SD) for Prader-Willi syndrome (PWS) deletion and uniparental maternal disomy (UPD) subjects, and equivalent controls

	Matching characteristic				
Subject group	**Full-scale IQ**	**Verbal IQ**	**Performance IQ**	**Age (years)**	**Fat (%)***
PWS deletion (*n* = 24)	62.7 ± 9.8	62.1 ± 9.5	66.5 ± 9.6	22.4 ± 8.3	51.2 ± 6.6
PWS UPD (*n*= 19)	63.9 ± 6.4	69.7 ± 5.5	61.6 ± 7.7	24.4 ± 9.3	47.2 ± 7.5
Equivalent control (*n* = 22)	71.5 ± 12.5	71.9 ± 11.2	75.4 ± 14.2	27.8 ± 13.1	48.3 ± 6.9

*Measured by Dean-energy X-ray absorptiometry (DEXA; Mazess *et al.* 1990).

Table 2

Mean proportion (± SD) of correct discriminations of kinematograms by Prader-Willi syndrome (PWS) deletion and uniparental maternal disomy (UPD) subjects, and equivalent and normal controls

	Kinematogram stimulus			
Subject group	**Correlation fixed, reduced density**	**Density fixed, reduced correlation**	**Density and correlation reduced**	**Total**
PWS deletion (*n* = 24)	0.86 ± 0.18	0.48 ± 0.15	0.61 ± 0.20	0.66 ± 0.14
PWS UPD (*n* = 19)	0.51 ± 0.38	0.27 ± 0.21	0.36 ± 0.30	0.38 ± 0.27
Equivalent control (*n* = 22)	0.82 ± 0.25	0.38 ± 0.21	0.48 ± 0.29	0.59 ± 0.27
Normal control (*n* = 14)	1.00 ± 0.00	0.54 ± 0.19	0.80 ± 0.17	0.78 ± 0.10