WHY HASN'T STUDYING PERCEPTION IN AUTISM SPECTRUM DISORDERS HELPED US CREATE A COGNITIVE MODEL?

Jonathan Adlam
Emmanuel College Cambridge and Clinical School University of Cambridge, Cambridge, UK

SUMMARY
There are a number of cognitive models of autism that aim to explain how mental processes are handled differently in the condition. These models make claims about the nature of cognitive function in people with autism, and suggest that these differences applied in social contexts lead to the characteristic behavioural patterns. However, it is difficult to study these cognitive differences directly because of the complexity of social situations. Studies of perceptual function are tempting as an alternative way to study cognition because it is far easier to control the conditions and the stimuli that participants are exposed to. This makes hypothesis generation and interpretation of results more objective and more convincing.

However, the study of perception in autism hasn't been very productive in contributing towards a model of cognition in autism. In many areas there are studies reporting contradictory results, preventing arrival at a consensus about the largest unresolved issues in the area. These studies tend to be repeated multiple times, but continue to provide contradictory evidence that doesn't allow us to place confidence in any of the cognitive models. An approach to these issues is proposed, focusing on critical analysis of contradictory studies rather than the endless process of repetition. This allows previous studies to be interpreted more objectively and resolve conflicts, and guides the design of future studies in ways that avoid the pitfalls that have been identified. Both of these outcomes result in more productive work being done.

The first example is in the study of motion perception in autism, where the use of non-identical stimuli has been problematic. On closer critical analysis, a fundamental aspect of the motion stimuli demonstrates that the contradictions might be expected based on the differences in stimuli used. Addressing this issue can move the field towards resolution. A second example is in the study of spatial frequency sensitivity. Here, poor study design has created results leading to an "eagle-eyed visual acuity" hypothesis of autism. Errors in the initial study are revealed, suggesting that the model should be abandoned. Finally, a general issue is the assumption of homogeneity of perceptual ability and genetics in autism, where the reality is that subgroups exist within the population of people with autism, and significant variation exists between them. The evidence for this is summarised and the issues that it creates explored.

Key words: autism spectrum disorders - perceptual ability - cognitive model - critical analysis

* * * * *

INTRODUCTION
There are a number of cognitive models of autism that aim to explain how mental processes are handled differently in the condition. They include the weak central coherence theory and the enhanced perceptual function theory. These models make claims about the nature of cognitive function in people with autism, and suggest that these differences applied in social contexts lead to the characteristic behavioural patterns. However it is difficult to study these cognitive differences directly. Social situations are complex and there are too many factors involved in behaviour that make these experiments hard to design, difficult to carry out, and almost impossible to interpret objectively. For these reasons and more, alternative methods are required to investigate cognitive function in people with autism, discriminate between the models that have been proposed, and decide whether they are accurate at all or whether new models need to be contemplated.

One alternative method is to study perceptual function in people with autism. Studies of perceptual function are tempting in this area because they are relatively simple and less costly, but most importantly it is far easier to control the conditions and the stimuli that participants are exposed to, making hypothesis generation and interpretation of results more objective and more convincing. There are a few fundamental assumptions that need to be laid on the table for this approach's strengths and weaknesses to be fully appreciated. It is important to note that the cognitive models of autism aim to explain the characteristic features, which tend to be associated with social situations. The cognitive processes associated with perception do not necessarily share the same differences as those associated with social behaviour. For the study of perception to be relevant to social behaviour, we must assume either that the cognitive differences in perception are similar to the cognitive differences in social behaviour, or that a perceptual difference in people with autism is somehow altering social behaviour. This isn't necessarily the case - it is very plausible that there is a social cognitive difference in autism, but that perceptual cognition would be entirely unrelated. It is also assumed that different performance in perceptual tasks in autism are due to different cognitive patterns. This is also not necessarily true, since performance is affected by factors such as intelligence, attention, and visual function. We can attempt to control for intelligence and visual function, but attention remains a problem in perceptual tasks.
With these limitations noted, the study of perception should provide at least some insight into cognitive patterns in autism and shed some light on the relevant cognitive models of autism. However, this hasn’t been achieved yet. In many areas there are studies reporting contradictory results, preventing arrival at a consensus about the largest unresolved issues in the area. These studies tend to be repeated in various forms, but continue to provide evidence that doesn’t allow us to gain confidence in any of the cognitive models. The way to solve this problem is not to conduct more studies that will be interpreted by both sides of the discussion as supporting their theory, but to critically analyse areas where there have been contradictory results, in an attempt to show why they are contradictory. If this is achieved, it will either allow previous studies to be interpreted more objectively and resolve conflicts, or it will allow future studies to be designed in ways that avoid the pitfalls that have been identified, and result in more productive work being done. There are several areas where critical analysis can shed light on the source of contradictory studies, demonstrating how this process should proceed.

THE USE OF DIFFERENT STIMULI BETWEEN STUDIES

Unfortunately, even in studies that use the same experimental paradigm, research groups in different laboratories have not shown exactly the same stimuli to their participants. This means that their results are not always entirely comparable. When there are contradictory results, careful analysis of the specific stimuli used is required. Subtly different stimuli might give results that look contradictory, while upon further investigation the differences in the stimuli can be informative themselves.

Case study: global motion integration

In the study of motion perception in autism, one area of contention is global motion processing. This is commonly tested using a ‘random dot kinematogram’, consisting of a field of moving dots. A certain proportion of these dots move together in one direction ("coherently"), while the rest move randomly. The subject needs to indicate in what direction the dots as a group seem to be moving. The test is used to extract a motion coherence threshold, which is the proportion of dots that need to move coherently to produce a target level of success (Simmons 2009).

It is assumed that the main strategy used to complete this perceptual task is to combine motion information from across the display. Effectively, 100 dots might be viewed, their direction averaged, and the output is perceived as the overall direction. This unconscious process, named global motion processing, represents the participant's ability to use integrated information from throughout the visual field to decide on the direction of motion, rather than using individual dots. The main alternative to using global motion information is to focus on the direction of an individual dot. To prevent this strategy being useful, the dots are given a limited lifespan, making this strategy quite ineffective.

This type of study could potentially be very informative about underlying cognitive differences in autism. If the weak central coherence model is correct, we might expect that the inability of autistic people to see the big picture will impair their performance on this task. They will not be as good at integrating motion information from across the display, which has been identified as the most effective strategy. However if the enhanced perceptual function model is correct, the ability to integrate information from across the display is intact, and the enhanced low-level perceptual ability will result in equivalent or better performance in the autistic group.

A number of studies have used paradigms of this kind, yet they draw contradictory conclusions about the ability of people with autism to use these global motion cues. Spencer et al. (2000), Milne et al. (2002), and Pellicano et al. (2005) concluded that individuals with autism have higher motion coherence thresholds, meaning that they are worse at integrating motion information from across the display. Similarly, Spencer and O'Brien (2006), Tsermentseli et al. (2008), and Milne et al. (2006) found that only subgroups of the autism spectrum, such as high-functioning autism, had higher motion coherence thresholds. These studies can be used as evidence in favour of the weak central coherence model. Finding contradictory results to these, Del Viva et al. (2006) did not observe any significant difference between their autism group and controls for three different types of motion. This would be consistent with the enhanced perceptual function model, and potentially problematic for the weak central coherence model.

Del Viva et al. (2006) and Pellicano et al. (2005) were described as the "most extreme" positions in a review on vision in autism (Simmons et al. 2009). They point out that although both used the same fundamental motion coherence paradigm, the specific stimuli used in each were different. In Del Viva et al. (2006), dots were larger and of more than one colour, as well as moving more rapidly and having a longer individual lifetime. It is suggested in Simmons et al. (2009) that these two results are not in fact contradictory, but instead represent an increased vulnerability in autism to correspondence noise, an unintended epiphenomenon of the experimental stimulus.

Correspondence noise is a term used by Barlow and Tripathy (1997) to describe the uncertainty present in random dot kinematograms. The stimuli themselves do not contain actual motion, but rather create the illusion of apparent motion between successive frames where each stationary dot has a slightly different position. In order to create the perception of motion, the visual system needs to decide which dot corresponds to which in successive frames. Since the dots are identical, this is
ambiguos and requires cognitive effort to decide which dots to pair together. Barlow and Tripathy (1997) outlined several parameters of kinematograms that can influence the amount of correspondence noise present in the display. Just one example is whether the dots are all the same colour or of many different colours. The colour of a dot is a very useful property when resolving the ambiguity because dots of different colours cannot be paired together. Having differently coloured dot populations decreases the number of possible dot correspondences and dramatically reduces the correspondence noise in the stimulus. A similar argument is made for whether the dots are all the same size, or vary in size. Other factors such as stimulus area alter correspondence noise by a smaller amount, but still make a contribution to the total amount of noise.

The stimulus of Del Viva et al. (2006) has lower correspondence noise mainly due to their use of dots of more than one colour. If people with autism have difficulty solving the correspondence problem using low-level perceptual ability, people with autism might need to use additional, higher-level cognitive abilities to make their choice. This would be expected to manifest as increased motion coherence thresholds in the autistic population since the unforeseen factor of correspondence noise will limit success in autistic subjects, rather than their ability to use global motion cues. If this is correct, the motion coherence thresholds of Del Viva et al. (2006) should be more accurate since the level of correspondence noise in their stimuli was lower, supporting the claim that global motion integration is not impaired in autism.

Conclusions

This instructive example demonstrates the need to critically evaluate the paradigms used in the field of studying perception in autism. Paradigms that allow unintended aspects of the visual stimulus to distort results should be discarded or refined to reduce this tendency. If not, their results will inevitably be difficult to interpret, hindering arrival at consensus.

STUDY DESIGN

There are a number of ways that a study's methodology might invalidate its results and conclusions. Almost all aspects of study design can influence the results, including statistical methods, equipment choice, and subject selection including diagnosis of autism. The choice of visual stimulus is of particular importance when studying perceptual ability. In a number of areas, there are examples of studies that use entirely inappropriate stimuli to measure perceptual abilities, and this shows itself in the contradictory results gained in studies of the same aspect of perception.

Using good experimental design not only reduces the chance of arriving at incorrect conclusions, but also increases the ease with which future researchers can resolve any contradictions between studies. Studies that simply compare a perceptual ability between autism and control groups make much more general predictions than one that is able to segment that ability into its component parts and make specific predictions about each.

Case study: spatial frequency sensitivity

Tasks that favour the use of local details require the processing of high-frequency visual information. Several researchers have investigated the perception of spatial frequency in autism in order to clarify whether well known phenomena such as increased autistic ability in tasks such as Embedded Figures is because of greater sensitivity to high spatial frequencies. This test requires the subject to pick out a simple shape from a more detailed, distracting stimulus. Since details are best encoded in high spatial frequency filters, and the big picture in low spatial frequency filters, subjects with the greatest sensitivity to high spatial frequencies are expected to be more proficient at finding the target shape within the distractions. Greater spatial frequency sensitivity would be consistent with the enhanced perceptual function model, while normal or reduced spatial frequency might prove problematic for the model.

An approach to studying this area is using visual acuity. These studies include de Jonge et al. (2007), which used a Landholt-C gap test, a clinical measure of visual acuity. Milne et al. (2009) used the Crowded Log MAR test, another clinical measure. Neither of these studies reported a significant difference in spatial frequency sensitivity between groups. A later study by Ashwin et al. (2009) using the Landholt-C stimuli reported abnormally high visual acuity in autistic subjects, an effect entirely absent from the earlier studies. This lead to their "eagle-eyed acuity" hypothesis, which is that individuals with autism have extremely high visual acuity that they calculated as approaching values reported in birds of prey. This visual acuity is interpreted as giving people with autism their generally exceptional performance on tasks such as the embedded figures test.

In resolving this contradiction, it is important to look at study design, and in this regard Ashwin et al. (2009) has been severely criticised. In practical terms, for the distance subjects were sat it has been claimed that the display was physically unable to present the spatial frequencies quoted as an upper limit for autistic subjects' visual acuity. This would mean that the study is simply unable to substantiate claims of such high visual acuity, and the results must be inaccurate or at least have been misinterpreted. Bölte et al. (2012) was aiming to replicate Ashwin et al.’s results in a more convincing way. They made improvements in the study design by using the same software, a digital version of Landolt-C, but utilised more standard parameters to generate stimuli, which should solve the resolution problems. They also obtained a larger sample size, and included an extra control group of schizophrenic
Conclusions

Clearly, well-designed studies are less liable to produce contradictory results. There are two particularly important experimental design points that can be taken from the example of spatial frequency selectivity. Firstly, clinical tests are not a suitable tool for determining whether there are small differences between groups in the context of a study. They are not sensitive enough to detect small differences in visual acuity. Since autistic people do not necessarily suffer from a visual condition, any difference in visual acuity between the autistic group and controls is unlikely to be as substantial as the impairments seen in clinical patients. The proposed perceptual difference therefore requires more sensitive tests. Koh et al. (2010) improved the field by using luminance static Gabor patches, which present a very restricted range of spatial frequencies to the eye. Other types of cue present a broad range of spatial frequencies, and allow subjects to use off-frequency visual cues to complete the task instead of the intended frequency. Therefore the use of Gabor stimuli gives more confidence that the aspect of perception being measured is in fact a particular spatial frequency.

Models of heterogeneity between individuals

Despite calculating significant heritability for autism of at least 0.9 (Rutter 1999), genome-wide association studies have failed to reliably find individual genes providing autism vulnerability. An example is the serotonin transporter gene, about which there have been inconsistent results from association studies (Kluck 1997). This suggests that there is no single causative gene for the disorder, and can be explained in a number of ways.

One explanation proposes that autism is highly polygenic and single genes have very small individual influences on the development of autism, with the disorder only developing when a number of predisposing genes are present. An example of this is the liability threshold (LT) model, which may be relevant in many psychiatric, polygenic conditions (Falconer 1965). Heterogeneity under the LT model would involve significant genetic differences between all individuals, perhaps to the extent that no two people with autism (excluding patients. The experimenters calculated that, given the effect size reported in Ashwin et al. (2009), their new test had extremely high power to detect that difference if it was correct. Despite this, they showed no significant difference between the groups. This forms a convincing rebuttal of the claims in Ashwin et al. (2009) about the extreme ability of the autistic group to detect higher spatial frequencies, and highlights the utility of critically reviewing previous studies, rather than repeating the same experiments with the same flaws.

The validity of Ashwin et al.’s statistical extrapolation of low visual acuity data to the upper threshold of visual acuity is also questioned (Crewther & Sutherland, 2009). Koh et al. (2010) tested a more extensive range of spatial frequencies, eliminating the need to extrapolate into the higher spatial frequencies. They used a staircase procedure with variable step size and sufficient repetition to ensure valid results.

The choice of visual stimulus is also suspect in Ashwin et al. (2009). All of the previously mentioned studies have all used clinical tests to measure spatial frequency sensitivity, designed for use as convenient and quick measures in diagnosis. They are not sensitive enough to detect small differences in visual acuity. Since autistic people do not necessarily suffer from a visual condition, any difference in visual acuity between the autistic group and controls is unlikely to be as substantial as the impairments seen in clinical patients. The proposed perceptual difference therefore requires more sensitive tests. Koh et al. (2010) improved the field by using luminance static Gabor patches, which present a very restricted range of spatial frequencies to the eye. Other types of cue present a broad range of spatial frequencies, and allow subjects to use off-frequency visual cues to complete the task instead of the intended frequency. Therefore the use of Gabor stimuli gives more confidence that the aspect of perception being measured is in fact a particular spatial frequency.

HETEROGENEITY OF AUTISM

Introduction

The assumption is often made that the population with autism is homogenous, so that any perceptual abnormality discovered will be replicable. Unfortunately, the population is not homogenous at either the genetic or phenotypic level, shown by study into the genetic differences between autistic people and the frequent failure of replication in certain perceptual areas. The variability that is apparent within the population means that perceptual ability varies between individuals, and complicates the process of identifying abnormalities and telling them apart from possible attentional differences.

Studying the sources of variability in autism has been productive in some ways, with examples of studies showing perceptual differences between classes of diagnosis on the autism spectrum, such as high functioning autism and Asperger's Syndrome (Spencer 2006). However, the existence of heterogeneity might be used in an unproductive way when failure to replicate is attributed to heterogeneity without specifying a measure that accounts for the difference. The major issue is that without an understanding of the structure of this heterogeneity, the implications of a null result explained by appeal to heterogeneity, or of a significant result within a particular autism spectrum subgroup, are unclear. Clarification of the structure of the variable autistic population will allow claims of heterogeneity to be scrutinised in a more methodical way, and guide better design of future studies based around this knowledge.
monzygotic twins) have exactly the same genetic influences on their condition. It proposes the existence of a set of genes that normally contribute to a healthily developing brain, where increased numbers of genetic knockouts results in movement along the autistic spectrum until a cut-off point where autism is arbitrarily defined for clinical and research purposes. The specific genetic knockouts will differ between individuals.

The second explanation proposes that different subgroups of autism have different genes providing vulnerability to autism. Here this is termed the subgroup homogeneity (SH) model. Heterogeneity under the SH model would involve varying genotypes across the spectrum, but contain discrete subgroups within which individuals have similar genetic aetiology. The two models are not mutually exclusive, but it is possible to investigate which is a better fitting model of the heterogeneity in autism by defining subgroups and seeing to what extent we can find genotypic uniformity within them. If we do find subgroups with significant uniformity, the SH model is probably a better way to describe genetic variability in autism. If not, it might be better to model autism as a single, highly polygenic disorder, using models such as LT.

Determining which model is more accurate has practical importance. In the study of perceptual differences, it will guide future study design, since the model of genetic heterogeneity that is accepted will determine what approaches are most productive. For example, under the SH model, studying subgroups on their own rather than the entire spectrum as a whole might be a more promising approach. At first glance this is odd since the normal strategy is to increase sample size when trying to improve statistical power, and studying subgroups will inevitably involve smaller samples. But if genetic causes are unique to subgroups, then studying the spectrum as a whole is counterproductive since only within subgroups will genetic similarities be found. On the other hand, under the LT model we would prefer to keep using genome-wide scans of the entire spectrum, with ever increasing sample sizes, as the best approach to finding any common genetic causes.

Evidence for uniform subgroups

There is a body of evidence suggesting that certain subgroups have greater genetic uniformity than the spectrum as a whole, supporting the SU model. In Buxbaum et al. (2001) a potential susceptibility locus on chromosome 2q was identified. The data for their whole sample were compared with the data for a restricted group, those with coincident autism and phase-speech delay (PSD). The linkage for the 2q region was a lot higher in the PSD group compared to the entire sample. As such, this subgroup was genetically distinguishable from other subgroups, and around chromosome 2q had greater within-group uniformity, precisely the findings that would be predicted by the SU model. This result suggested that the 2q region contains a set of genes that is particularly relevant for the development of coincident autism and PSD, but the region is not as relevant in the development of other subtypes of autism. This can explain why other studies may have concluded that this locus is not as important as it appeared here, since for the majority of people with autism without PSD this region is not linked as strongly to the development of the disorder. These data do not appear to be consistent with the LT model of autism, which would expect the linkage in the subgroup to approximate the linkage of the population as a whole. For this reason, their data suggest that genetic causes differ between their chosen subgroup and the spectrum as a whole. Other subgroups of this type are likely to exist, with separate genetic aetiology.

There is also some support for genetic uniformity within other subtypes, including Asperger's Syndrome (Ylisaukko-oja 2004), this particular study finding a locus for Asperger's Syndrome susceptibility. These results need to be replicated in other subgroups of autism in order to demonstrate that studying subgroups is a promising technique. On the other hand, some subgroups that have been studied showed little genetic homogeneity. An example is in Rehnström et al. (2009) where the Finnish population, considered to be a highly genetically homogenous group, displayed extremely variable genetic risk factors with very little overlap. Clearly the Finnish population cannot be considered as one of these genetically homogenous subgroups. Evidence is required before any particular subgroup can be treated as one with a common genetic cause.

Conclusions

Two models of heterogeneity in autism have been presented that aim to describe the genetic variation between individuals: the subgroup uniformity model and the liability threshold model. The discovery of subgroups showing higher genotypic uniformity than the spectrum as a whole suggests that the SH model makes useful distinctions between groups. This means that future studies into perceptual abnormalities in autism should ideally attempt to investigate subgroups of autism, not only the spectrum as a whole. This approach has already been taken up by many of researchers, who aim to study a category such as Asperger's syndrome or high-functioning autism. However, these two broad categories are not necessarily the ones that best account for genotypic variation in autism, and the choice of subgroups should in future be guided by the results of investigation into the genetic structure of subgroups. The coincident autism and PSD group used in Buxbaum et al. (2001) would most likely be missed if the categories studied were broad ones such as Asperger's syndrome.

Within the field of perception, the aim is to elucidate the genetic causes and implications of perceptual differences. In this context, until genetically distinct groups are identified, it makes sense to subdivide the
spectrum by perceptual ability. Within such subgroups, there might be greater phenotypic uniformity and so study results might be more easily replicated. The categories of Asperger's Syndrome and high-functioning autism are not specified by perceptual ability, and so their use in this field is minimal. Instead, researchers might consider dividing the spectrum by perceptual ability. For example, in studying global motion coherence, a group of subjects could be chosen who perform within a certain range on a test of central coherence, such as the embedded figures task. This would select a subgroup of those with, say, weak or strong central coherence. Then their motion coherence thresholds can be compared to controls and to the autism spectrum as a whole. This might provide a platform for a more rational and productive investigation into the weak central coherence explanation for perceptual differences, giving the evidence that different subgroups have important differences. Positive results found in this way could identify subgroups for study.

**DISCUSSION**

Studying perception provides a practical way to investigate the cognitive processes of people with autism. The simplicity and ability to control experimental variables means that these studies have a huge advantage over other forms of research that aim to achieve the same outcome. However, the history of the field is that contradictory results can be found, allowing both sides of the discussion to cite them in their favour, and hindering any real progress from being made. Three major factors contributing to this situation are non-uniformity of stimuli, erroneous study design, and the assumption of homogeneity among an autistic population that is actually complicated and heterogeneous. The study of perceptual differences in autism will only create useful results if studies are designed properly, heterogeneity is taken into account, and research groups aim to use identical stimuli when repeating studies or designing new ones. The critical analysis of previous studies will allow conflicts to be explored further, leading towards either resolution or providing clarity on how to come to it via novel investigations.

**Acknowledgements:** None.

**Conflict of interest:** None to declare.

**References**