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Discussion forum

Improving diagnosis of developmental prosopagnosia: The role of exclusion criteria

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Recently there is renewed interest in the diagnosis of developmental prosopagnosia (DP) (Burns, 2024; Burns et al., 2023; DeGutis & Campbell, 2024; DeGutis et al., 2023; Gerlach et al., 2024; Lowes et al., 2024). Several topics have been discussed, including how much prevalence rates of DP vary depending on inclusion cut-offs (DeGutis et al., 2023), whether self-report data should weigh more than objective test scores (Burns et al., 2023; 2024), how correlation between tests can bias prevalence estimates (Gerlach et al., 2024), and how response time data can improve diagnostic sensitivity (Lowes et al., 2024). Here we add a discussion about exclusion criteria, which can be used to rule out alternative explanations for face recognition deficits. We focus on two criteria for which we have collected data, namely mid-level visual deficits (i.e., trouble with intermediate visual processes that link "low-level" image representations and "high-level" interpretations of objects and scenes) and autism traits, and we estimate their prevalence in a sample of 1479 people who self-referred to us for DP diagnosis online. Depending on inclusion criteria, 7–11% of DPs may present with mid-level visual deficits and 14–21% with substantial autism traits. These estimates suggest that face recognition deficits in a non-trivial proportion of DPs may result from issues or involve dysfunctions beyond face processing impairments.

We start by describing our sample demographics. Our 1479 participants comprise 1078 females, 363 males, and 38 others. The age range is 18–77 years ($M = 35.89$, $SD = 9.25$). All statistical tests we report below use uncorrected p-values. Participants completed three standard inclusion measures for DP: Prosopagnosia Index 20 (PI20) (Shah et al., 2015), Cambridge Face Memory Test (CFMT) (Duchaine & Nakayama, 2006), and a famous face test (FFT) (Duchaine & Nakayama, 2005). As expected, all three measures correlate with each other, demonstrating the assessment of the same construct (PI20-$CFMT_{accuracy}$ $r = -.208$, $p < .001$; PI20-$FFT_{accuracy}$ $r = -.195$, $p < .001$; $CFMT_{accuracy}$ – $FFT_{accuracy}$ $r = .484$, $p < .001$). For diagnostic thresholds we use the norm from the PI20 original report (score >64, Shah et al., 2015) and in-house data for CFMT ($M = 78\%$, $SD = 11\%$) and FFT ($M = 79\%$, $SD = 12\%$). Using PI20 as the sole inclusion criterion (i.e., PI20 score >64, Shah et al., 2015), 96% of the sample (1420/1479) can be classified as "self-report DP". Using the DSM-5 framework for mild or major neurocognitive disorders (i.e., PI20 score >64 plus CFMT and FFT z-scores of −1 or below, DeGutis et al., 2023; Sachdev et al., 2014), 64% of the sample (947/1479) meet the criteria for "mild or major DP". Using the most popular diagnostic approach (i.e., PI20 score >64 plus CFMT and FFT z-scores of −2 or below, DeGutis et al., 2023), 35% of the participants (513/1479) meet the threshold for "classic DP". Our estimates of 96%, 64%, and 35% accord with previous corresponding estimates of 100%, 62% and 38% from Bate et al. (2019; $N = 165$), and of 100%, 70%, and 44% from Burns (2023; $N = 61$). Our larger sample size bolsters past estimates and suggests that

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these statistics likely reflect the underlying characteristics of self-referring DP samples.

While inclusion measures are essential because they establish the presence of face recognition problems in DP, exclusion measures are important because they help rule out alternative explanations for DP that involve factors beyond face processing (Barton & Corrow, 2016; Dalrymple & Palermo, 2016). Given the hierarchical nature of the human visual system, a traditional exclusion criterion is low-level visual impairment, commonly assessed with tasks such as judgments of orientation, size, and length from the Birmingham Object Recognition Battery (Riddoch & Humphreys, 1993), as well as assessments of visual acuity and contrast sensitivity. DPs rarely have trouble with such tasks (e.g., Bate et al., 2019; Behrmann et al., 2005; Garrido et al., 2008; Lowes et al., 2024), leading to the suggestion that low-level assessment in DP might be excessive (Burns, 2024).[1] However, newer and potentially more sensitive low-level tests are available (e.g., Kieseler et al., 2022), and they might be able to pick out subtle deficits that would have gone undetected.

Less often examined but is also relevant to DP is a range of intermediate or mid-level visual functions. These include figure-ground segmentation, perceptual grouping, local and global processing, and fine shape perception (Torfs et al., 2014). Mid-level visual deficits have been observed in multiple conditions including visual agnosia (Milner et al., 1991), simultanagnosia (Robertson et al., 1997), neglect (Driver & Mattingley, 1998), autism (Dakin & Frith, 2005), schizophrenia (Silverstein & Keane, 2011), and acquired prosopagnosia (Sergent & Signoret, 1992; Rezlescu et al., 2012; Kosslyn et al., 1995; Barton et al., 2004). Mid-level vision has not been thoroughly examined in DP, but several studies using minimal or degraded stimuli provide relevant data. For example, one study found abnormal performance when DPs had to detect two-tone faces with missing contours that are embedded among similar two-tone distractors, which presumably requires some degree of perceptual grouping, visual closure, or both (Garrido et al., 2008). Some DPs also had difficulties identifying objects at the basic level, especially when the object images are fragmented (Gerlach et al., 2016). In contrast, two studies found intact perception of Mooney faces in DP, one using a simple intact versus scrambled discrimination task (Le Grand et al., 2006) and the other an ERP adaptation task (Towler et al., 2016). Together, these studies indicate that some aspects of mid-level vision can be atypical in some DPs and are therefore worth assessing.

We assessed mid-level vision with The Leuven Perceptual Organization Screening Test (L-POST) (Torfs et al., 2014), a clinically-oriented assessment of a range of mid-level visual function. L-POST consists of 15 subtests, each testing a particular mid-level function or process. Scores under 10th percentile on 4 or more subtests are taken to indicate potential deficits (Torfs et al., 2014). Across the whole sample, L-POST correlates with CFMT ($r = -.17$, $p < .001$) and FFT ($r = -.09$, $p < .001$) but not PI20 ($r = -.01$, $p = .786$). Depending on inclusion criteria, our data show that 7–11% of DPs may have mid-level vision issues (96/1420 of self-report DPs, 79/947 of mild/major DPs, and 58/513 of classic DPs). This analysis shows that a non-trivial percentage of DPs may have mid-level vision problems, which can account for part of their face recognition deficits and may reflect more extensive visual problems beyond face processing. Moving forward, we recommend researchers to use mid-level visual deficits as an exclusion criterion, which we and others have done (Bell et al., 2023; DeGutis et al., 2024; Fry et al., 2020, 2023; Little et al., 2022; Little & Susilo, 2023; Mishra et al., 2021; Smith & Susilo, 2021). Screening for mid-level vision might also obviate the need to assess low-level vision, given the reasonable assumption that participants with serious low-level problems would likely present with mid-level deficits.[2]

Another common exclusion criterion is autism. Although autism and DP can doubly dissociate and are likely to be independent (Duchaine et al., 2009; Fry et al., 2023; Kamensek et al., 2023), the two conditions are related. Face recognition deficits are a common feature of autism (Griffin et al., 2021; Hedley et al., 2011; Minio-Paluello et al., 2020; Weigelt et al., 2012), and autism traits can be elevated in DP (Bate et al., 2019; Bell et al., 2023; Fry et al., 2023). In the typical population, autism traits have been associated with face recognition skills, although the evidence is mixed (Halliday et al., 2014; Lewis et al., 2018; Verhallen et al., 2017). The relationship between face recognition deficits and autism traits in DP is complex. More research is needed to clarify whether autism traits can account for face recognition deficits in DP, and whether face recognition deficits can account for the presence of autism traits in DP.

We assessed autism traits using Subthreshold Autism Trait Questionnaire (SATQ), a 24-item self-report scale that measures a broad range of subthreshold autism tendencies in the general population (Kanne et al., 2012). Across the whole sample, SATQ correlates with CFMT ($r = -.15$, $p < .001$), FFT ($r = -.12$, $p < .001$), and PI20 ($r = .06$, $p = .02$). A SATQ score of >40 is taken to index the presence of significant autism traits (Kanne et al., 2012). Depending on inclusion criteria, between 14 and 21% of DPs qualify as having significant autism traits (202/1420 of self-report DPs, 160/947 of mild/major DPs, and 107/513 of classic DPs). These estimates might be somewhat inflated since two SATQ items explicitly address perception of face expression (e.g., "I am good at knowing what others are feeling by watching their facial expressions or listening to the tone of their voice"), which can be affected in DP (Bell et al., 2023; Biotti & Cook, 2016; Djouab et al., 2020; Burns et al., 2017; Palermo et al., 2011; Tsantani et al., 2022). However, many DPs have normal expression perception (Bell et al., 2023;

[1] There might be a file drawer problem here since studies could have excluded DPs with low-level deficits without reporting it.

[2] It is possible that mid-level deficits may interact with profiles of DP. For example, DPs with mid-level deficits may be more likely to show impairments with non-face objects compared to DPs without. For such studies, researchers may want to include mid-level functions in the main analysis rather than using them to exclude participants. The challenge is to demonstrate that such mid-level deficits have a specific relationship with the characteristics of the high-level deficits with faces or objects (e.g., trouble with curvature perception would selectively impact faces or other "curvy" objects). The mid-level deficits must also not explain away DP since they might produce face-level impairments when the face mechanisms themselves are actually intact.

Dobel et al., 2007; Duchaine et al., 2003; Humpreys et al., 2007), and where expression deficits occur, they are less severe compared to identity deficits (Bell et al., 2023; Djouab et al., 2020) and are linked to autism traits (Bell et al., 2023; Fry et al., 2023).

Should autism traits be used as an exclusion criterion? We think not, at least in general. Our recommendation is to assess autism traits whenever possible and consider them in the main analysis. This approach can help determine features of DP that are related to autism and those that are not, which can improve our knowledge of both DP and autism. This is what we did recently to reveal the link between autism traits and expression deficits but not identity deficits in DP (Bell et al., 2023). More generally, this strategy also fits within the transdiagnostic framework, an approach for studying psychological and psychiatric conditions that cut across traditional diagnostic domains, aiming to yield general insights that apply beyond specific populations (Astle & Fletcher-Watson, 2020). However, there can be exceptions. For example, researchers may want to study aspects of DP which we know are characteristic of autism, such as communication difficulties (e.g., maintaining eye contact). In this case, excluding participants based on autism traits may make sense. Another example is a resource-intensive neuroimaging study with a limited sample size. Researchers may choose to exclude participants based on autism traits to increase the likelihood of the sample having similar neural abnormalities.

In terms of specific measures, we chose SATQ because it was designed to capture a broader range of autism traits compared to other measures such as the Autism Quotient (AQ) or the Broad Autism Phenotype Questionnaire (BAPQ). However, SATQ, AQ, and BAPQ all correlate strongly (range .73–.79 in a student sample and .63–.83 in autism sample; Kanne et al., 2012), so any of them would be appropriate for indexing the general presence of autism traits within DP. However, some measures may be more relevant for some research questions depending on the items they include. For example, SATQ has items on expressive language, eye contact, and odd behaviours, whereas AQ does not. Where exclusion is desirable, we suggest a practical cut-off score of 40+ for SATQ, which is comparable to a cut-off score of 32+ for AQ (Kanne et al., 2012).

Other exclusion criteria beyond mid-level visual deficits and autism traits have been used or proposed, among others abnormal intelligence (Bate et al., 2019; Fry et al., 2023), person recognition deficits (Gainotti, 2013), structural lesion in MRI scans (Avidan et al., 2005; Liu et al., 2015), and broader recognition problems as assessed with words (Corrow et al., 2016) or objects (Gerlach et al., 2024). Each of these criteria has merit, but it is not practical to use them all for every study. Moreover, different criteria would be more or less valuable depending on research questions. For example, a study focusing on the social consequences of face recognition failures may not need to exclude participants with object recognition problems, whereas a study of face-selective mechanisms in DP would need to. It seems prudent to distinguish between necessary exclusions to increase sample homogeneity and optional exclusions tailored to specific research aims. To this point, we hope to encourage researchers to be more careful in their choice of exclusion measures and to start a discussion about mandatory exclusion criteria that we as a field can agree upon. As a starter, we recommend (1) low-level or mid-level visual deficits and (2) known history of brain damage or injury.

These are exciting times in DP research. Getting DP diagnosis right would go a long way in moving our field forward. Real and important issues must be discussed and debated, and we have much to gain from exchanging views. But we also need to agree on some diagnostic common ground, or else it would be difficult to know whether we are studying the same thing. Lurking behind this challenge is the ontological problem of what "DP" means (Barton & Corrow, 2016). As a field, we need to decide whether DP has a particular meaning that refers to some underlying cause (e.g., face recognition deficits due to impairments in face processing mechanisms) or to some common statistical threshold for atypical performance (e.g., face recognition skills worse than 95% of the population), as opposed to being a catch-all label for anyone who thinks they have bad face recognition. Ontological problems are not unique to DP – any psychological condition or psychiatric disorder that can be viewed as dimensional or categorical or must grapple with it (Haslam et al., 2020). The problem will probably not be resolved soon, but we can make progress by collecting more empirical data and assessing various diagnostic criteria more comprehensively.[3]

CRediT authorship contribution statement

Tirta Susilo: Writing – review & editing, Writing – original draft, Conceptualization. **Brad Duchaine:** Writing – review & editing.

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[3] A promising approach for future studies of DP diagnosis is taxometrics, which offers statistical tools to assess whether a psychological condition is typologically distinct from normal variability (Haslam et al., 2020; Meehl & Golden, 1982). Recent DP studies have started to use related tools such as cluster analyses (Bennetts et al., 2022; DeGutis et al., 2023, 2024), but taxometric methods provide several advantages (Beauchaine, 2007).

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