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A family at risk: Congenital prosopagnosia, poor face recognition and visuoperceptual deficits within one family

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ABSTRACT

Congenital prosopagnosia (CP) describes a severe face processing impairment despite intact early vision and in the absence of overt brain damage. CP is assumed to be present from birth and often transmitted within families. Previous studies reported conflicting findings regarding associated deficits in nonface visuoperceptual tasks. However, diagnostic criteria for CP significantly differed between studies, impeding conclusions on the heterogeneity of the impairment. Following current suggestions for clinical diagnoses of CP, we administered standardized tests for face processing, a self-report questionnaire and general visual processing tests to an extended family ($N=28$), in which many members reported difficulties with face recognition. This allowed us to assess the degree of heterogeneity of the deficit within a large sample of suspected CPs of similar genetic and environmental background. (a) We found evidence for a severe face processing deficit but intact nonface visuoperceptual skills in three family members – a father and his two sons – who fulfilled conservative criteria for a CP diagnosis on standardized tests and a self-report questionnaire, thus corroborating findings of familial transmissions of CP. (b) Face processing performance of the remaining family members was also significantly below the mean of the general population, suggesting that face processing impairments are transmitted as a continuous trait rather than in a dichotomous all-or-nothing fashion. (c) Self-rating scores of face recognition showed acceptable correlations with standardized tests, suggesting this method as a viable screening procedure for CP diagnoses. (d) Finally, some family members revealed severe impairments in general visual processing and nonface visual memory tasks either in conjunction with face perception deficits or as an isolated impairment. This finding may indicate an elevated risk for more general visuoperceptual deficits in families with prosopagnosic members.

1. Introduction

Individual recognition of familiar faces is one of the most important and demanding abilities for humans in social life (e.g., Farah, Wilson, Drain, & Tanaka, 1998; Young, De Haan, & Bauer, 2008). The very high performance in this skill is assumed to be subserved by cortical networks specialized on the processing of faces (e.g., Haxby, Hoffman, & Gobbini, 2000; Kanwisher, McDermott, & Chun, 1997). Lesions within these cortical networks can lead to a state in which patients are dramatically impaired in recognizing faces, despite normal lower-level vision, object identification skills, and semantic knowledge. This severe neurological impairment has been called *prosopagnosia* or *face blindness* and has attracted a lot of interest in the last decades, both in the scientific community and in the general population. Prosopagnosia provides evidence that face processing is a cognitive function that may be dissociated from general visual processing or object processing (e.g., Farah, 1996; Moscovitch, Winocur, & Behrmann, 1997).

1.1. Congenital prosopagnosia

Individuals with an isolated face recognition deficit, which manifests itself in early childhood but is not attributable to overt neurological, neuropsychological, or psychiatric abnormalities, have been categorized as congenital, developmental, or hereditary prosopagnosics (e.g., Behrmann & Avidan, 2005; Duchaine & Nakayama, 2006b; Jones & Tranel, 2001; Kennerknecht, Grüter,

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Welling, & Wentzek, 2006; Kress & Daum, 2003). In line with our earlier publications, we will use the term congenital prosopagnosia (CP) to emphasize its presumed presence from birth and/or its hereditary origin. A significant number of case descriptions, but also group- and family studies on this condition have been published in recent years (e.g., Dobel, Bolte, Aicher, & Schweinberger, 2007; Duchaine, Germine, & Nakayama, 2007a; Grueter et al., 2007; Kennerknecht, Ho, & Wong, 2008a; Kennerknecht, Pluempe, & Welling, 2008b; Kennerknecht, Pluempe, Edwards, & Raman, 2007; Schmalzl, Palermo, & Coltheart, 2008).

Independent research groups have estimated the prevalence of CP at 2–3% in the general population (Bowles et al., 2009; Kennerknecht et al., 2006, 2008a). However, it is not clear whether these findings indicate a dichotomous, bimodal distribution of face processing skills in the population, or whether CPs reflect the lower end of a normal distribution, which would imply a continuous representation of face processing skills in the general population. Recent evidence of so-called *super recognizers* who perform approximately 2 SDs above the mean of the general population, but also population-based assessments with sensitive behavioral tests point towards the latter interpretation of the results (Kennerknecht, Kischka, Stemper, Elze, & Stollhoff, 2011; Russell, Duchaine, & Nakayama, 2009; Wilmer, Germine, Chabris, Gerbasi, & Nakayama, 2012).

Given that many CPs report on first-degree relatives who are also impaired in face recognition, most researchers argue for a hereditary contribution to CP (Behrmann & Avidan, 2005; De Haan, 1999; Dobel et al., 2007; Galaburda & Duchaine, 2003; Kennerknecht et al., 2008b). Support for a genetic contribution to face recognition skills in the general population arises from studies in behavioral genetics (Wilmer et al., 2010; Zhu et al., 2010). These studies have compared performance of mono- and dizygotic twins on different tests of face cognition and have estimated the specific impact of genetic variation in face recognition to be as high as 39%. Such findings imply that CP and face recognition deficits more generally may be predominantly found in certain families.

Regarding the underlying cognitive mechanism of the impairment, there is some evidence that persons with CP display abnormalities in what is called *configural* or *holistic* processing of faces (e.g, Avidan, Tanzer, & Behrmann, 2011; Palermo et al., 2011; Robbins & McKone, 2007; Tanaka & Farah, 1993; Van Belle, De Graef, Verfaillie, Busigny, & Rossion, 2010). Usually people perceive an upright face as an indecomposable whole, despite its constitution of individual features with complex spatial relations to each other (Maurer, Grand, & Mondloch, 2002). Behavioral evidence for this special cognitive treatment of upright faces comes most prominently, from the *face inversion effect* (Yin, 1969). The *face inversion effect* describes reduced recognition rates for faces that are presented upside down (i.e., inverted) compared to upright faces. This disproportion is considerably larger for faces compared with other objects. Supposedly, the inversion of a face as well as a misalignment of the bottom half (*composite face effect*, e.g., Young, Hellawell, & Hay, 1987) interferes with the interactive processing of its parts, leading to a feature-based, analytic encoding strategy, which is less efficient than a holistic approach regarding accurate and fast recognition. In prosopagnosic subjects however, these usually robust behavioral effects are often not found; in fact, many described cases even display better recognition rates for inverted faces (Avidan et al., 2011; Busigny & Rossion, 2011; Dobel, Putsche, Zwitserlood, & Junghofer, 2008; Duchaine, Yovel, & Nakayama, 2007b; Farah, Wilson, Drain, & Tanaka, 1995; Lee, Duchaine, Wilson, & Nakayama, 2010; Schmalzl, Palermo, Harris, & Coltheart, 2009).

1.2. Heterogeneity of CP as a clinical condition

Whereas an impairment of face recognition is by definition at the core of CP, evidence on associated neuropsychological deficits in persons classified as CP is scattered and often conflicting, suggesting that CP may be a heterogeneous clinical condition (Dobel et al., 2007; Le Grand et al., 2006; Schmalzl et al., 2008). Among the reported perceptual deficits in nonface visual domains are intraclass object agnosia (i.e., difficulties in discriminating between members of other semantic categories such as houses or cars; Behrmann, Avidan, Marotta, & Kimchi, 2005; Duchaine et al., 2007b), impaired perception of biological motion including lip reading (Dobel et al., 2007; Lange et al., 2009), and visual imagery deficits (Tree & Wilkie, 2010). In other cases, however, the face recognition deficit was reported to be isolated, or at least not associated with deficits in object processing or domain-general visual abilities (e.g., Duchaine & Nakayama, 2005; Stollhoff, Jost, Elze, & Kennerknecht, 2011).

One cause of this heterogeneity regarding associated deficits may be genuine diversity in the investigated cases themselves (i.e., subjects may present with various subtypes of CP depending on genetic and/or environmental factors). A second cause for the reported heterogeneity may be a poor comparability between the employed methods and diagnostic criteria to classify individuals as CP. Such methods range from self-reports (e.g., Dinkelacker et al., 2010; Grueter et al., 2007; Kennerknecht et al., 2006) to interpreting significant group differences in tailor-made experimental tasks (e.g., Behrmann et al., 2005; Dobel et al., 2007; Duchaine & Nakayama, 2005; Le Grand et al., 2006). Moreover, among the latter types of studies the tested domains and neuropsychological functions vary considerably in their level of specificity and difficulty, ranging from low-level face perception of gender or emotion to highly abstract and difficult tasks on nonface perceptual organization. From a practitioner's point of view, basic research on CP has suffered from a lack of consensus on clear criteria both for diagnosis and for exclusion (Gainotti, 2010; Herzmann & Danthiir, 2008). However, the situation has been improved by Bowles et al. (2009), who suggested consensual clinical diagnostic criteria based on neuropsychological face processing tests which provide normative data, cut-off scores and a high level of psychometric quality.

With this study, we attempt to contribute to the current debate on heterogeneity of CP as a clinical entity. We analyzed the patterns of performance on face processing tasks and general visual processing tasks across a large family sample ($N=28$) in which many members reported face recognition difficulties. The studied sample is highly similar with regard to genetic and environmental factors, especially within core families (e.g., a father and his offspring). This high group homogeneity allows us to largely control for the impact of genetic and environmental variability on performance. Previous work on CP within family samples concluded that CP is a primarily heterogeneous condition regarding associated neuropsychological deficits and/or underlying cognitive functions (Lee et al., 2010; Schmalzl et al., 2008). We were now interested in whether the use of the recently suggested standardized diagnostic criteria and clinical cut-off scores (Bowles et al., 2009) might yield a more homogeneous picture of the condition within families by reducing the possibility of falsely diagnosing CP e.g., in ambiguous cases. For single-case diagnoses, such a normative account has several advantages over a group comparison of tailor-made experimental tasks. First of all, normative samples are usually larger, allowing for a more precise quantitative classification of the results compared to smaller control group samples. Second, standardized tasks have usually been tested for their psychometric quality. Most importantly, published and standardized neuropsychological tests provide a basis for reproducible results and comparisons between studies and thus constitute the standard procedure in clinical settings.

With this normative account, we furthermore aimed to contribute to the current debates on whether CP is a categorical or a