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Research report

Three cases of developmental prosopagnosia from one family: Detailed neuropsychological and psychophysical investigation of face processing

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A B S T R A C T

A number of reports have documented that developmental prosopagnosia (DP) can run in families, but the locus of the deficits in those cases remains unclear. We investigated the perceptual basis of three cases of DP from one family (67 year-old father FA, and two daughters, 39 year-old D1 and 34 year-old D2) by combining neuropsychological and psychophysical methods. Neuropsychological tests involving natural facial images demonstrated significant face recognition deficits in the three family members. All three members showed normal facial expression recognition and face detection, and two of them (D2, FA) performed well on within-class object recognition tasks. These individuals were then examined in a series of psychophysical experiments. Intermediate form vision preceding face perception was assessed with radial frequency (RF) patterns. Normal discrimination of RF patterns in these individuals indicates that their face recognition difficulties are higher in the cortical form vision hierarchy than the locus of contour shape processing. Psychophysical experiments requiring discrimination and memory for synthetic faces aimed to quantify their face processing abilities and systematically examine the representation of facial geometry across viewpoints. D1 showed deficits in perceiving geometric information from the face at a given view. D2's impairments seem to arise in later face processing stages involving transferring view-dependent descriptions into a view-invariant representation. FA performed poorly on face learning and recognition relative to the age-appropriate controls. These cases provide evidence for familial transmission of high-level visual recognition deficits with normal intermediate-level form vision.

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1. Introduction

The present study provides a detailed investigation of three cases of developmental prosopagnosia (DP) from one family. Prosopagnosia is a neurological condition characterized by an impairment in face recognition. While acquired prosopagnosia is caused by a brain damage, DP is manifested in the absence of any discernible brain lesion and neuro-developmental disorders (e.g., Asperger syndrome) (Behrmann and Avidan, 2005; Kress and Daum, 2003). Recently, multiple cases of DP in the same family have been reported (Duchaine et al., 2007a; Grueter et al., 2007; Schmalzl et al., 2008), suggesting the heritability of this syndrome (McConachie, 1976). However, little is known about the perceptual basis of these apparently inherited cases of prosopagnosia. In the present study, three of five family members across two generations showed significant deficits in face processing despite normal visual sensory and intellectual function. The affected individuals were then tested with a series of psychophysical experiments that were designed to identify the perceptual locus of the face processing deficits.

Face processing consists of a number of hierarchical stages and parallel processes in a distributed cortical network (Bruce and Young, 1986; Haxby et al., 2000) (see Fig. 1). Thus, the present study used psychophysical tests that systematically assessed different face processing stages. To date, most research with familial prosopagnosics has only assessed early vision and higher-level processes. However, face processing deficits could result from a problem in any part of the network including mid-level visual processes. To evaluate the possibility of general perceptual deficits at the level of intermediate form processing, perception of closed curvature in the DP participants was examined using radial frequency (RF) patterns (Wilkinson et al., 1998). In earlier studies, most DP individuals have performed normally with intermediate form vision tasks involving concentric Glass patterns (Le Grand et al., 2006) or Navon letters (Duchaine et al., 2007a, 2007b; but also see Behrmann et al., 2005; Bentin et al., 2007). Glass pattern detection measures sensitivity to structure in global form, requiring integration of local elements into a global configuration (Gallant et al., 1996; Wilson et al., 1997). The Navon task assesses global-local perception using compound letter stimuli (Navon, 1977). However, neither task involves closed contour curvature, likely a direct input to face processing mechanisms (Wilkinson et al., 2000; Wilson et al., 2000). RF patterns used in the present study are comprised of curvatures and circles that are key attributes of faces and may most effectively probe intermediate form vision important to face and object perception (Wilkinson et al., 1998, 2000). In a functional magnetic resonance imaging (fMRI) study, concentric patterns produced activation similar to the level elicited by faces in V4 and half as much activation as faces in the fusiform face area (FFA) (Wilkinson et al., 2000). In addition, evidence from psychophysical and fMRI data suggests that analysis of concentric patterns in V4 contributes to face processing (Wilkinson et al., 2000; Wilson et al., 2000).

We also used psychophysical tests to examine what types of face processing operations are deficient in the three DPs. Face processing deficits could result from a difficulty in

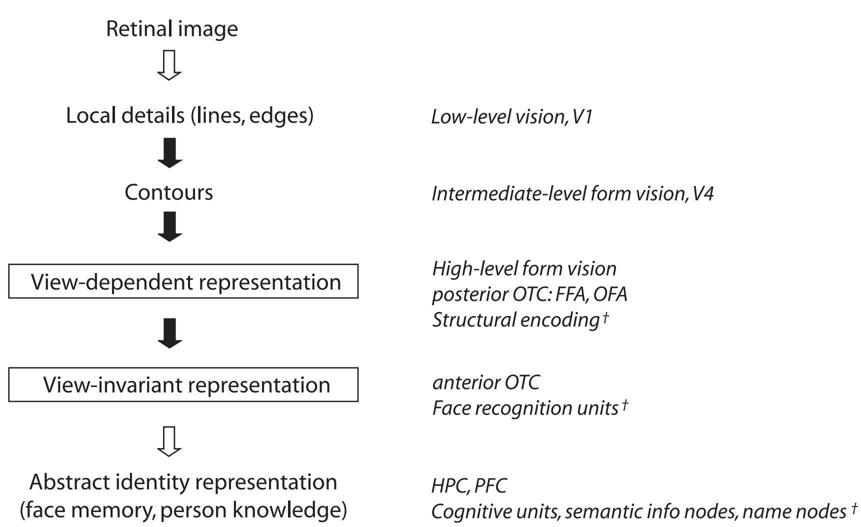

Fig. 1 – A schematic diagram of face processing stages in the brain. Visual representation of faces involves multiple cortical regions along the hierarchy of the ventral visual pathway. Fine representation of individual faces at a particular view would be formed in the (FFA, Kanwisher et al., 1997) and occipital face area (OFA, Gauthier et al., 2000) (Andrews and Ewbank, 2004; Grill-Spector and Malach, 2001). View-invariant representation of facial identity, across a large change in viewpoint, would be achieved in later brain regions (Eger et al., 2005; Pourtois et al., 2005a, 2005b) by associating disparate view representations (Riesenhuber and Poggio, 1999; Wallis and Bülthoff, 2001). Face learning and memory would involve the hippocampus (HPC) and prefrontal cortex (PFC) (Haxby et al., 1996; Quiroga et al., 2005). In real life, faces are hardly ever encountered from identical vantage points. But in an experimental situation where faces are learned and recognized in the same view, view-invariant representation stage could be bypassed. There is not only the feed-forward processing but also top-down modulation to support view-invariant representation and learning (Riesenhuber and Poggio, 1999). The data from DP individuals suggest that connectivity among functional systems is a vital component of face analysis (e.g., Thomas et al., 2009). †face processing stages in Bruce and Young (1986). OTC = occipito-temporal cortex.