 American Journal of Medical Genetics Part A 146A:2860–2862 (2008)

Invited Comment

Comment on Prevalence of Hereditary Prosopagnosia (HPA) in Hong Kong Chinese Population

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Received 16 August 2008; Accepted 27 August 2008

Kennerknecht et al. [Kennerknecht et al. (2008); Am J Med Genet Part A 146A] estimate that 1.9% of the Chinese population are hereditary prosopagnosics. I discuss concerns about their assumption that the great majority of prosopagnosia resulting from developmental problems are heritable and present data from my laboratory that suggests that a considerable proportion of developmental prosopagnosics do not have relatives who share their face recognition deficits. © 2008 Wiley-Liss, Inc.

Key words: prosopagnosia; developmental prosopagnosia; cognitive genetics; neuropsychology

How to cite this article: Duchaine B. 2008. Comment on prevalence of hereditary prosopagnosia (HPA) in Hong Kong Chinese population. Am J Med Genet Part A 146A:2860–2862.

Reports of severe selective deficits affecting face recognition first appeared in the neurological literature in the mid-19th century [Wigan, 1844; Jackson, 1876; Quaglino et al., 2003]. These individual had intact early visual processes and normal intelligence but had lost the abilities necessary to compute identity from the face. This striking condition—called *prosopagnosia*—usually caused serious social difficulties for patients experiencing it [Bornstein, 1963; Yardley et al., in press]. For years, prosopagnosia was thought to occur only after brain damage, but it is clear now that many people are prosopagnosic not because of damage to mechanisms involved in face recognition but because of a failure to develop those mechanisms. These developmental prosopagnosics (sometimes called *congenital*) experience similar difficulties as people who acquire their prosopagnosia later in life. A number of laboratories are investigating the cognitive and neural basis of developmental prosopagnosia [for reviews, see Behrmann and Avidan, 2005; Duchaine and Nakayama, 2006], but the developmental nature of the condition obviously raises the question of whether it is caused by genetic factors. Hints that developmental prosopagnosia was sometimes due to genetic factors have been present since the first case report [McConachie, 1976]. Within recent years, a number of articles have reported families with multiple prosopagnosics [De Haan, 1999; Duchaine et al., 2003; Behrmann et al., 2005; Kennerknecht et al., 2006], including articles with behavioral data documenting prosopagnosia in families with 10 [Duchaine et al., 2007] and seven [Schmalzl et al., 2008] affected members.

In this issue, Kennerknecht et al. follow up their previous estimate of the prevalence of hereditary prosopagnosia in a German sample by investigating its prevalence among Han Chinese. In the German study, they estimated that 2.5% of Germans are hereditary prosopagnosics [Kennerknecht et al., 2006, 2007]. I question the classification of all of the prosopagnosics in that sample as hereditary cases and it will be essential to support the self-report data by which they classified prosopagnosic cases with behavioral testing, but their estimate suggests that developmental prosopagnosia is not a rare disorder. My experience suggests this estimate is quite plausible. After I present talks on prosopagnosia, I'm regularly approached by audience members who tell me that they or someone close to them is prosopagnosic. My laboratory has been contacted by more than 3,000 self-reported prosopagnosics through our web page (http://www.faceblind.org), and when articles or programs on prosopagnosia are presented in major media outlets, we are contacted by hundreds of prosopagnosics. In addition to their

Grant sponsor: ESRC; Grant number: RES-061-23-0400.

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Published online 16 October 2008 in Wiley InterScience (www.interscience.wiley.com)

DOI 10.1002/ajmg.a.32548

scientific value, the estimates by Kennerknecht et al. also have been important in raising awareness of the condition for the public (and hopefully for funding agencies!) through media coverage of their articles.

In the current article, they used a two-step process to identify prosopagnosics in a group of 533 medical students at the University of Hong Kong. The students first filled out a screening questionnaire. Of the 133 students with responses deemed typical of prosopagnosia, 120 agreed to take part in a semi-structured interview lasting approximately 1 hr. In it, the students were asked about their medical histories, strategies they use to deal with their face recognition problem, anecdotes indicative of prosopagnosia, and whether any relatives shared their difficulties.

Ten students were classified as prosopagnosic, and four of the 10 reported family members with prosopagnosia. Based on their finding of 10 students out of 533, Kennerknecht et al. [2008] estimated the prevalence of hereditary prosopagnosia to be 1.9%. Note however that only four of the prosopagnosics reported family members with prosopagnosia. The authors do not discuss their rationale for classifying the six prosopagnosics without affected relatives as hereditary cases. This inference is problematic for two reasons—one theoretical and one empirical.

This study was motivated by the possibility that the prevalence of hereditary prosopagnosia among Han Chinese might differ from that of ethnic Germans. Given the size of the Han Chinese population, this certainly seems worth investigating and it also provides another opportunity to estimate the prevalence of prosopagnosia. The authors' categorization of the six as heritable suggests that they assume that the proportion of hereditary to non-hereditary prosopagnosics does not vary between populations. However, this assumption is theoretically questionable. If the prevalence of prosopagnosia may vary between groups, then the proportion of different types of prosopagnosia may vary as well.

I am also skeptical of the classification of the six prosopagnosics as heritable, because my laboratory works with a considerable number of developmental prosopagnosics who are unaware of family members with face recognition difficulties [Duchaine and Nakayama, 2005]. To provide an estimate of the heritable and non-heritable cases who visit my lab, consider 19 participants involved in a combined behavioral and neuroimaging study that we are in the midst of. Each of these participants reports regular difficulties with face recognition in everyday life and has scored significantly worse than controls on a number of face memory tasks. A radiologist has inspected their structural scans and reported that none have observable lesions so their face recognition problems appear to result from developmental problems. Of these 19 participants, 11 report close relatives with face recognition problems, four are unsure, and four are confident that none of their relatives are prosopagnosic (see Fig. 1). It is possible that some of these participants are unaware of family members who are impaired with face recognition but these prosopagnosics have asked their relatives about their face recognition abilities so it seems very unlikely that all have relatives with prosopagnosia.

Given these considerations, it seems prudent that only the four Chinese participants who reported family members should contribute to the estimate. This would revise the estimate from 1.9% to 0.75%, but it still suggests that a staggering number of Han Chinese have hereditary face recognition problems. With a population of 1.3 billion worldwide, 0.75% translates to 9.75 million Chinese hereditary prosopagnosics. In addition, the estimate of 1.9% for the prevalence of developmental prosopagnosia in the Chinese population is valuable in that it provides further evidence that this condition is common and merits greater attention.

Acknowledgement that some prosopagnosics are not hereditary in origin is important for at least two reasons. First, the presence of such prosopagnosics highlights the importance of first separating hereditary prosopagnosics from apparently non-hereditary cases in studies attempting to identify the genetic basis of prosopagnosia so that the non-hereditary cases do not add noise to the results. Second, suggestions that prosopagnosia is always heritable would lead some prosopagnosics without affected relatives to conclude that they do not meet the criteria for prosopagnosia. This would be unfortunate because many people with face recognition deficits find great relief in the realization that their difficulties are due to problems with perception or visual memory. Many prosopagnosics have told me that

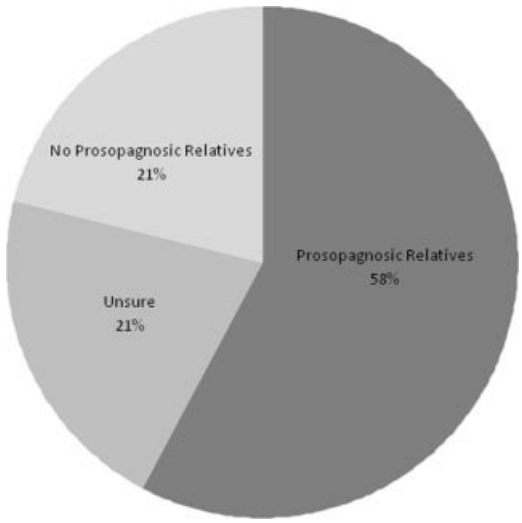

FIG. 1. Family histories in a group of developmental prosopagnosics. Nineteen developmental prosopagnosics tested in my laboratory reported whether genetic relatives also have significant face recognition difficulties. Eleven reported affected relatives, four were unsure, and four were confident that none of their relatives shared their face recognition difficulties.

they felt guilty until they learned about the condition because they thought that they were not trying hard enough or simply did not care enough about others to remember faces.

ACKNOWLEDGMENTS

This work was supported by an ESRC grant (RES-061-23-0400). Thanks to Lucia Garrido for providing the structural imaging results and for editorial suggestions.

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