Am. J. Hum. Genet. 62:221–223, 1998

INVITED EDITORIAL
Instant Recognition: The Genetics of Pitch Perception

Peter K. Gregersen

Division of Biology and Human Genetics, North Shore University Hospital–New York University School of Medicine, Manhasset, NY

For most of us, quick and accurate perception of the visual world is essential for getting around in life; we take for granted our instant recognition of color, shape, distance, and the physical relationships between objects. Many aspects of the auditory world are also apprehended in an effortless manner: a nighttime whistle is easily identified as a distant train, without any deliberation. However, identifying the pitch of an isolated whistle is beyond the abilities of most people. This is also the case for most musicians, despite the fact that they spend every day working in the context of a standardized system of pitch relationships. Those rare individuals who can instantly recognize the pitch of a random piano tone or passing car horn, without the use of a reference pitch, possess a cognitive ability that is termed "absolute pitch" (AP), also called "perfect pitch" in the vernacular.

Pitch is a one-dimensional attribute defined by the number of vibrations, per second, emanating from a sound source, such as a plucked string (Rasch and Plomp 1982). In reality, a plucked string vibrates at many different frequencies, with wavelengths that correspond to the entire length of the string and to fractions thereof (½, ⅔, ¼, etc.). Vibrations with these fractional wavelengths produce pitches that are termed "overtones." Nevertheless, the casual listener usually perceives the fundamental frequency (with a wavelength of the entire string) as being *the* pitch of the note being played. The mixture and timing of the overtones are responsible for the characteristic sound quality, or timbre, of the particular instrument (Risset and Wessel 1982). The range of useful musical pitches is ~20–5,000 Hz, which is, roughly, the range of a piano keyboard. "Concert A" is defined as a pitch with a vibration frequency of 440 Hz. In practice, local standards for concert A may vary in different parts of the world (European orchestras often tune slightly higher), which can be distracting for some musicians who possess AP.

The peripheral auditory organs are designed specifically for frequency analysis. The cochlear basilar membrane vibrates, at each point along its length, with an optimal resonant frequency. The fact that all humans are quick to appreciate the differences in timbre between instruments illustrates the extreme sensitivity of this organ to complex frequency spectra. Therefore, AP perception is not dependent on a special kind of ear; it reflects a particular ability to analyze frequency information, presumably involving high-level cortical processing (Zatorre and Beckett 1989; Zatorre et al. 1994; Schlaug et al. 1995).

During the last century, scientific opinion about the etiology of AP has been spread widely across the "nature-nurture" continuum (Ward and Burns 1982). Many studies have suffered from low statistical power, a lack of controls, and a minimal appreciation for the complexities of genetic causation. The report by Baharloo et al. (1998 [in this issue]), with Nelson Freimer and his colleagues at the University of California San Francisco (UCSF), marks the dawn of a refreshing new era in this regard. By surveying and testing a large population of musicians, the authors have begun to generate basic information on the prevalence and familial aggregation of AP and on environmental factors that influence the development of AP. Particularly important is the careful attention paid by Baharloo et al. to defining the phenotype. Although AP is qualitatively distinct from the relative-pitch ability possessed by all trained musicians, it nevertheless exhibits some phenotypic heterogeneity, and this is likely to be an important consideration for successful mapping studies.

Some aspects of the results reported by Baharloo et al. (1998) are surprising. Of 612 musicians returning surveys, fully 15% reported AP. This compares with previous undocumented estimates of <1/1,500 amateur music students (Profita and Bidder 1988), and it is well above our own results of ~1%–2% of several hundred music students and professionals (Gregersen and Kumar 1996). The reason for this discrepancy is unlikely to be inaccurate self-reporting, since AP ability is quite distinctive, and objective testing confirms that the majority

Received December 10, 1997; accepted for publication December 11, 1997; electronically published January 30, 1998.

Address for correspondence and reprints: Dr. Peter K. Gregersen, Division of Biology and Human Genetics, North Shore University Hospital, NYU School of Medicine, 350 Community Drive, Manhasset, NY 11030. E-mail: peterg@nshs.edu

This article represents the opinion of the author and has not been peer reviewed.

0002-9297/98/6202-0004$02.00

of self-reporters indeed have AP. Possessors of AP may be more likely to respond to surveys. Alternatively, the particular group of musicians surveyed may have been partially preselected for AP. This could result either from having attended certain music schools or from attaining a particular kind of musical proficiency demanded by the high-quality musical organizations that were surveyed. In Japan, for example, AP ability is specifically valued in musicians, and very young children may receive training directed toward their developing AP. Although no control data are available, a careful reading of the Japanese literature suggests that the majority of students in Japanese music conservatories possess at least some degree of AP (Miyazaki 1988); presumably, this is due to selection in the admissions process. Clearly, additional surveys will be required, to determine the prevalence of AP in various populations.

A second major finding of the UCSF group is that musicians with AP tend to start their musical education quite early in childhood, nearly all at <6 years of age. This result is interpreted as providing support for the hypothesis that early exposure to music, beginning perhaps at a so-called critical time during development, is necessary but not sufficient for the development of AP. However, as the authors acknowledge, it is also possible that the presence of AP in a child may lead to displays of interest in music, which in turn provoke parents to provide music lessons early. Genes and environment interact, and the interaction goes both ways. Teasing apart these effects, especially in retrospect, is extremely difficult, and it is wise to be cautious about drawing any firm conclusions from this kind of historical data.

Nevertheless, other evidence suggests that childhood exposure has an impact on the AP phenotype. The careful studies of Miyazaki (1988) and of others (Takeuchi and Hulse 1991) have shown that some pitches are identified more accurately than others, by many AP possessors. In particular, accidentals (the black notes on the piano keyboard) are recognized less easily than the white notes. Early childhood music education commonly begins with melodies in keys that contain few accidentals, and this may explain the bias toward accurate recognition of tones in the C major scale.

Although Profita and Bidder (1988) reported familial aggregation of the AP phenotype, their study was uncontrolled in that it was without a rigorous determination of background prevalence. We have recently estimated the relative risk to sibs (λ_s) to be ~20 (Gregersen and Kumar 1996). The data of Baharloo et al. (1998) on the affected-sibling rate are controlled for early music education in the siblings, and these data suggest an λ_s of ~7.5 (95% confidence interval of 2.2–21.2), by our calculation. While twin data for a relatively rare phenotype such as AP are difficult to generate, we have ascertained three sets of identical twins with AP, all concordant, compared with one set of DZ twins who are discordant. Overall, the data strongly indicate that a substantial genetic component underlies the development of AP.

Since possession of AP is clearly not a prerequisite for outstanding musicianship, it is reasonable to ask about the broader practical significance of this phenotype. Is AP associated with other unusual cognitive abilities? Our informal impression is that a subset of AP possessors also exhibit a high degree of mathematical and memory ability and that, in rare instances, they may exhibit unusual perceptual talents in other sensory realms, such as taste. Perhaps it would be interesting to ask whether there is a higher prevalence of AP among professional wine tasters or their first-degree relatives! A small fraction of individuals with AP also experience strong color associations with particular pitches, a phenomenon that is termed "synesthesia." This was apparently the case among a number of composers, including Sibelius and Scriabin (Profita and Bidder 1988). Synesthesia is extremely rare in the general population, but, interestingly, it appears to exhibit strong familial aggregation (Baron-Cohen et al. 1996; Yoon 1997).

AP has also been reported anecdotally in the setting of certain neurological disorders, particularly autism and Williams syndrome (Sacks 1995), although the true prevalence has not been established with any degree of accuracy. Ascertainment of the AP phenotype in these disorders is extremely difficult, since testing requires that subjects possess a knowledge of music (i.e., knowing the common names of pitches). A proportion of individuals with Williams syndrome are gifted musically, even in the face of significant mental disability. However, a preliminary search for linkage between AP ability and genes in the Williams region on chromosome 7 has yielded negative results (Gregersen et al. 1997). The genetics of autism appears to be quite complex (Smalley 1997), but it is possible that there is genetic overlap between AP ability and autism, since some autistic individuals are musical savants who possess AP.

Ultimately, the real significance of AP may be that it is an unusually discreet and quantifiable cognitive phenotype that clearly has a substantial genetic component. This will make it possible to open a window on the relationship between inheritance, brain development, early childhood education, and cognition. The work of Baharloo and colleagues (1998) is an elegant prelude to what promises to be a crescendo of activity toward mapping and identifying the relevant genes.

References

Baharloo S, Johnston PA, Service SK, Gitschier J, Freimer NB (1998) Absolute pitch: an approach for identification of genetic and nongenetic components. Am J Hum Genet 62: 224–231 (in this issue)

Baron-Cohen S, Burt L, Smith-Laittan F, Harrison J, Bolton P (1996) Synaesthesia: prevalence and familiality. Perception 25:1073–1079

Gregersen PK, Kowalsky E, de Andrade M, Jawaheer D (1997) Affected sib pair analysis of families with absolute pitch (AP): exclusion of the Williams locus. Am J Hum Genet Suppl 61:A399

Gregersen PK, Kumar S (1996) The genetics of perfect pitch. Am J Hum Genet Suppl 59:A179

Miyazaki K (1988) Musical pitch identification by absolute pitch possessors. Percept Psychophysiol 44:501–512

Profita J, Bidder GT (1988) Perfect pitch. Am J Med Genet 29:763–771

Rasch RA, Plomp R (1982) The perception of musical tones. In: Deutsch D (ed) The psychology of music. Academic Press, New York, pp 1–24

Risset J-C, Wessel DL (1982) Exploration of timbre by analysis and synthesis. In: Deutsch D (ed) The psychology of music. Academic Press, New York, pp 26–58

Sacks O (1995) Musical ability. Science 268:621

Schlaug G, Jancke L, Huang Y, Steinmetz H (1995) In vivo evidence of structural brain asymmetry in musicians. Science 267:699–701

Smalley SL (1997) Genetic influences in childhood-onset psychiatric disorders: autism and attention-deficit/hyperactivity disorder. Am J Hum Genet 60:1276–1282

Takeuchi AH, Hulse SH (1991) Absolute-pitch judgments of black- and white-key pitches. Mus Percept 9:27–46

Ward WAD, Burns EM (1982) Absolute pitch. In: Deutsch D (ed) The psychology of music. Academic Press, New York, pp 431–451

Yoon CK (1997) Synesthesia: the taste of music, the sound of color. J NIH Res 9:25–27

Zatorre RJ, Beckett C (1989) Multiple coding strategies in the retention of musical tones by possessors of absolute pitch. Memory Cognit 17:582–589

Zatorre RJ, Evans AC, Meyer E (1994) Neural mechanisms underlying melodic pitch perception and memory for pitch. J Neurosci 14:1908–1919