

# Absolute Pitch: An Approach for Identification of Genetic and Nongenetic Components

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## Summary

Absolute pitch (AP) is the ability to recognize a pitch, without an external reference. By surveying more than 600 musicians in music conservatories, training programs, and orchestras, we have attempted to dissect the influences of early musical training and genetics on the development of this ability. Early musical training appears to be necessary but not sufficient for the development of AP. Forty percent of musicians who had begun training at  $\leq 4$  years of age reported AP, whereas only 3% of those who had initiated training at  $\geq 9$  years of age did so. Self-reported AP possessors were four times more likely to report another AP possessor in their families than were non-AP possessors. These data suggest that both early musical training and genetic predisposition are needed for the development of AP. We developed a simple computer-based acoustical test that has allowed us to subdivide AP possessors into distinct groups, on the basis of their performance. Investigation of individuals who performed extremely well on this test has already led us to identify several families that will be suitable for studies of the genetic basis of AP.

## Introduction

Absolute pitch (AP), also known as “perfect pitch,” refers to the ability to recognize the pitch of a musical tone, without an external reference pitch. There is general, although not universal, agreement that, to be considered an AP possessor, an individual must have the

ability to recognize pitches accurately and instantaneously (Takeuchi and Hulse 1993). Interpretation of the existing literature on AP is complicated by substantial variation in how AP has been operationally defined and tested. For example, some investigators have insisted that AP possessors be capable of producing specific tones, without reference either vocally or by use of a tone generator (Petran 1939; Revesz 1953), whereas others have focused only on the ability to recognize a pitch (Takeuchi and Hulse 1991). Similarly, some investigators have tested subjects for the ability to actually identify the pitch of tones with different timbres, such as sine-wave tones or piano tones, whereas others have limited testing to tones of a single timbre (Rakowski and Morawska-Bungeler 1987; Miyazaki 1989).

Although much is known about the anatomy and physiology of the human auditory pathway, the specific neural substrates involved in pitch perception remain unclear. Psychophysical and physiological experiments suggest that high-level cortical processes are involved in pitch perception (Klein et al. 1984; Zatorre et al. 1992, 1994). Recent positron-emission-tomography studies of musicians with and without AP indicate that anatomical asymmetry of the planum temporale (an associative auditory area of the brain) may be involved in the processing of pitch perception (Schlaug et al. 1995). Although these and other studies offer some information about the neurobiology of auditory perception, there is no evidence regarding underlying developmental mechanisms that may play a role in these processes. Isolation of genes responsible for AP could illuminate the developmental basis of pitch perception.

We hypothesize that the development of AP depends on both genetic and nongenetic influences. The genesis of neural circuits for many animal behavioral traits and attributes, such as the regulation of circadian activities (Takahashi 1996), follows a developmental blueprint that mostly is determined genetically. A number of studies suggest a genetic basis for AP (Revesz 1953; Bachem 1955). Recently, Profita and Bidder (1988) presented pedigrees with a high prevalence of AP. They suggested that, in these families, AP was inherited as an autosomal dominant trait with incomplete penetrance. There also

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is persuasive evidence that an environmental influence, in the form of very early musical training, contributes to the development of AP (Sergeant 1969; Miyazaki 1988a; Takeuchi 1989). Such studies suggest the existence of a so-called critical period for the genesis of AP; it is known that the development of certain neural circuits depends on the presence of sensory stimuli during such critical periods (Goodman and Shatz 1993). This proposed critical period for the development of AP may parallel the period during which children's speech perception becomes specialized for the sounds of their native language (Takeuchi and Hulse 1993).

Difficulties in the evaluation of evidence for both genetic and nongenetic influences on AP are that most previous studies have been conducted by use of small samples (Sergeant 1969; Miyazaki 1988a, 1988b; Profita and Bidder 1988; Takeuchi 1989) and that few studies have attempted to examine both kinds of influence. In this paper, we present data from a survey of a large sample of professional musicians and music students. The aim of the survey was to assess the role of musical training in the development of AP and to evaluate whether this trait aggregates in families. Our data support the hypothesis that both genetic and nongenetic factors contribute to the development of AP. The results of the survey encouraged us to develop an auditory test for AP, which we have used to initiate collection of a sample of AP possessors, for the purpose of conducting genetic mapping studies of AP.

## Subjects and Methods

### *Initial AP Survey*

To obtain background information on AP, we distributed a survey to musicians and music students in several music institutions and music performing groups. The survey was approved by the Committee on Human Research at the University of California San Francisco, and additional approval was obtained from the appropriate authorities at each institution or music performing group. A copy of the survey is available by request.

Nine hundred surveys were either distributed on site by the authors or sent by mail for distribution at the San Francisco Conservatory of Music, University of California San Francisco Symphony, Berkeley Music School at the University of California, Curtis Institute of Music, San Francisco Symphony Youth Orchestra, Peabody Conservatory of Music, La Scala Opera, Aspen Music School, and Interlochen Center for the Arts. Of these 900 surveys, 612 were completed and returned.

The text at the beginning of the survey explained the purpose of the survey and emphasized that, for our study, it was important to have all individuals respond, regardless of their AP status. This survey had three aims: (1) to determine the percentage of musicians in this sam-

ple who claim to possess AP; (2) to ascertain whether the age at first formal musical training correlates with the development of AP; and (3) to determine whether AP aggregates in families.

To determine whether the respondents possess AP, we inquired about their ability to identify the pitch of tones, in the absence of an initial reference. So that the responses of those who claimed AP were as objective as possible, we asked specific questions dealing with the speed and accuracy of pitch judgment, whether the subjects' AP depended on the tones generated by a particular instrument, and whether the subjects could vocally produce tones without first hearing a reference tone. To assess the role of musical training in the development of AP, we asked respondents about the extent of their musical training and their age at their first formal music lessons. In order to determine whether AP is aggregated in families, we asked the respondents about the presence of AP among their first-degree relatives. Associations between AP status and other variables were assessed by  $\chi^2$  contingency-table analyses or by Fisher's exact test.

### *Testing for AP and Follow-up Interviews*

Portable auditory tests were developed to assess AP ability in the self-reported AP possessors. Two types of tones were used as the stimuli for the tests: pure sine-wave tones and real piano tones. Digitized tones were stored on a portable computer (Apple 190) and were delivered to the subjects via headphones. We tested 48 musicians who claimed to possess AP and 12 musicians (sampled from music conservatories) who did not report AP.

*Stimuli.*—Sine-wave tones were digitally synthesized (16 bit, sampling rate 44.1 kHz) as text files on a Silicon Graphics Indy workstation by use of MatLab software (Mathworks) and were converted to standard Audio Interchange File Format audio files by use of conversion utilities. Tones of different frequencies were synthesized with different amplitudes, to equalize perceived loudness (as judged by S.B. and P.A.J.). Each tone had a duration of 1,000 ms, with onset and offset ramps of 100 ms. Sine-wave tones had frequencies corresponding to the 40 musical notes from C2 to G#8, on the basis of A4 = 440 Hz. Frequencies in the first octave were not used, since pure tones in this range were not reproduced clearly with our equipment.

Piano tones were taken from a CD produced by McGill University (Opolko and Wapnock 1987), containing professionally sampled tones from a 9-foot Steinway grand piano tuned to A4 = 440 Hz. The 40 piano tones from C1 to G#7 were digitally recorded from the CD to a MacIntosh PowerPC and were edited, by use of SoundEdit 16 software (Macromedia), to have uniform durations of 1,000 ms, with offset ramps of 100 ms. Half the tones presented to the subjects were equivalent

to those represented by the white keys on the piano, and half the tones were equivalent to those represented by the black keys. Tones from the eighth octave were not used, because of the insufficient duration of such high notes on the piano.

*Pitch-testing procedure.*—Two tests were administered to each subject. The first consisted of 40 pure tones and the second of 40 piano tones. These tests were divided into four blocks, with 10 trials in each block and 3-s intervals between each trial. Tones were played in pseudorandom order, with the constraint that successive tones were separated by more than two octaves and a semitone. Subjects listened to the tones through headphones and were asked to make an instantaneous judgment of the pitch of each tone and to write the name of the pitch on a sheet of paper. Subjects were not allowed any practice runs, and feedback regarding their performance was not given until the testing was completed.

*Analysis of test results.*—Responses to the auditory tests were scored in the following manner: all correct judgments were given one point. Because previous studies as well as anecdotal reports have suggested that AP is accurate to within a semitone (Baggaley 1974; Miyazaki 1988a), responses that were erroneous by one semitone were given  $\frac{3}{4}$  of a point, in order to enable us to distinguish between those AP possessors who make semitone errors and those who do not. Judgments that were more than a semitone from the actual pitch were given zero points. There have been anecdotal reports that, as the result of aging, an AP possessor's pitch perception may be shifted by a semitone (Ward and Burns 1982). In addition, of the 20 AP possessors tested who were >45 years of age, 16 reported that their pitch perception had shifted, by as much as a semitone, as they had become older. This claim was confirmed by the auditory tests of these individuals. Therefore, we decided to score a full point for semitone errors made by individuals >45 years of age. Tones at the extreme octaves (four pure tones in the eighth octave and four piano tones in the first octave) were excluded from scoring, since the performance of self-reported AP possessors and that of non-AP possessors was indistinguishable in these registers.

To determine categories of AP for future studies, we used the following procedure: 12 individuals who had similar levels of musical training and who were self-reported non-AP possessors were given the auditory test. Their pure and piano scores ( $x_{\text{pure}}$  and  $x_{\text{piano}}$ , respectively) were combined with the pure and piano scores from 12 randomly selected self-reported AP possessors, and the mean pure and piano scores and the standard errors (SEs) for these means were calculated. This procedure was repeated 100 times, and the means of these 100 means and SEs were calculated for the pure and piano

scores ( $\bar{x}_{\text{pure}}$ ,  $\overline{SE}_{\text{pure}}$  and  $\bar{x}_{\text{piano}}$ ,  $\overline{SE}_{\text{piano}}$ , respectively). The distribution of scores for this sample of musicians (self-reported AP possessors and self-reported non-AP possessors) was used to define categories of AP; individuals whose scores were most deviated above the mean were considered to have the most clear-cut AP. The categories are based on the assumption that accurate and instantaneous recognition of pure tones is the best indicator of AP. Thus, we based the definition of AP-1 (the category indicating clear-cut AP) only on the subjects' performance on the pure-tone tests, reasoning that they would be unlikely to attain a high score on this test if they needed to rely on external cues (e.g., timbre) and assuming that all individuals who scored well on the pure-tone tests also would score well on the piano-tone tests. We intended this category to include the individuals who would be probands in future genetic studies of AP. We also designed categories (AP-2 and AP-3) to include individuals with probable AP (on the basis of excellent but not outstanding scores on the pure-tone tests), as well as a category (AP-4) to include individuals whose pitch perception for pure tones is much worse than that of the individuals in the other three categories but who have outstanding pitch perception for piano tones. We reasoned that the AP-4 phenotype could have a basis different from those for the other three categories.

We determined the distribution of scores for the combined sample of self-reported AP possessors and self-reported non-AP possessors and established the four categories of AP (AP-1, AP-2, AP-3, and AP-4). The maximum score that could be obtained for each test was 36. The mean scores for this combined sample were 17.35 for pure tones and 20.24 for piano tones. Self-reported AP possessors whose score for pure tones was greater than  $\bar{x}_{\text{pure}} + 3 * (\overline{SE}_{\text{pure}})$  were designated as having AP-1 (i.e., a pure-tone score >24.49). Self-reported AP possessors whose score for pure tones was less than  $\bar{x}_{\text{pure}} + 3 * (\overline{SE}_{\text{pure}})$  but greater than  $\bar{x}_{\text{pure}} + 2 * (\overline{SE}_{\text{pure}})$  and whose score for piano tones was greater than  $\bar{x}_{\text{piano}} + 3 * (\overline{SE}_{\text{piano}})$  were designated as having AP-2 (i.e., a pure-tone score between 22.11 and 24.49 and a piano-tone score >27.79). Self-reported AP possessors whose score for pure tones was less than  $\bar{x}_{\text{pure}} + 3 * (\overline{SE}_{\text{pure}})$  but greater than  $\bar{x}_{\text{pure}} + 2 * (\overline{SE}_{\text{pure}})$  and whose score for piano tones was less than  $\bar{x}_{\text{piano}} + 3 * (\overline{SE}_{\text{piano}})$  were designated as having AP-3 (i.e., a pure-tone score between 22.11 and 24.49 and a piano-tone score <27.79). Self-reported AP possessors whose score for pure tones was less than  $\bar{x}_{\text{pure}} + 2 * (\overline{SE}_{\text{pure}})$  but whose score for piano tones was greater than  $\bar{x}_{\text{piano}} + 3 * (\overline{SE}_{\text{piano}})$  were designated as having AP-4 (i.e., a pure-tone score <22.11 and a piano-tone score >27.79). All other individuals were not assigned an AP designation. In our initial testing of the instrument, we tested 20 of the 92 self-reported AP possessors identified through the survey, as well as 28

self-reported AP possessors who came to our attention through word of mouth among musicians; these individuals were designated as having AP-1, AP-2, AP-3, or AP-4, or they were not assigned an AP designation. We subsequently tested an additional 51 self-reported AP possessors who came to our attention, via word of mouth, after the initial survey. On the basis of their scores, these individuals also were designated as having AP-1, AP-2, AP-3, or AP-4, or they were not assigned an AP designation.

#### Family and Musical History Interview

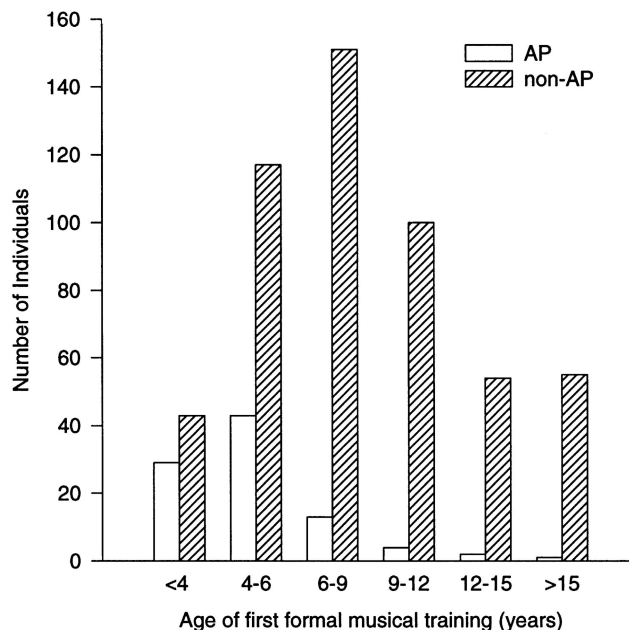
Subjects who participated in the testing for AP were also interviewed. Detailed questions regarding their musical training were asked, such as their exact age at their first music lessons, whether their early music lessons included any particular methods of ear training, and the reasons why they began music lessons when they did. A detailed pedigree of the family of each subject was drawn. For the first-degree relatives of each proband, the following information was obtained by report of the proband: the extent of musical training, the age at their first formal music lessons, and whether they possess AP. We performed auditory testing on those relatives of AP possessors who were readily available and who were self-described AP possessors.

## Results

### Initial AP Survey

In the population that we surveyed, the frequency of self-reported AP was 15% (92 of 612 respondents). There were no significant differences in the proportion of AP possessors among male and female respondents (data not shown). As shown in figure 1, 29 (40%) of 72 individuals who had begun their musical training at age <4 years reported AP. In contrast, 43 (27%) of 160 individuals who had received their first musical training at age 4–6 years and 13 (8%) of 161 individuals who had received their first musical training at age 6–9 years reported AP. Only 4 (4%) of 104 individuals who had begun musical training at age 9–12 years and 3 (2.7%) of 112 individuals who had begun musical training at age >12 years reported AP. These results indicate that there is a correlation between early musical training and the development of AP.

Previous studies have suggested that AP possessors may have the ability to vocally produce any tone, without a reference. In response to the question of whether the subjects are able to vocally produce any particular pitch without first hearing a reference tone, 85 (92%) of 92 self-reported AP possessors replied positively (we did not subsequently test for this ability). Other studies have suggested that AP ability may depend on the timbre



**Figure 1** Development of AP correlated with age at first formal musical training. The number of AP possessors and non-AP possessors is based on the self-reports of the survey respondents.

of a particular instrument with which individuals are most familiar. In our survey, most AP possessors (73 [79%] of 92) indicated that they could identify the pitch of tones produced by any instrument. However, the results of our testing indicate that timbre may be important for a subgroup of AP possessors.

To evaluate whether AP aggregates in families, subjects were asked to indicate whether they were aware of any family members whom they believed to be AP possessors. Of the self-reported AP possessors, 44 (48%) of 92 individuals indicated that they had first-degree relatives who also possessed AP. In contrast, only 72 (14%) of 520 non-AP possessors reported first-degree relatives with AP ( $\chi^2 = 38.6$ , 1 df,  $P < 10^{-5}$ ). Approximately equal proportions of self-reported AP possessors and non-AP possessors (28% and 32%, respectively) reported that they did not know whether any of their first-degree relatives possessed AP. These results suggest that AP is aggregated in families and may indicate that a genetic mechanism is involved in the development of AP.

To gain information regarding whether familial aggregation of AP may have a genetic basis, we performed an additional analysis of families in which both the survey respondent (regardless of whether he or she self-reported AP) and one or more siblings had received musical training at age <6 years. Of 15 siblings of self-reported AP possessors, who had received early musical training, 9 were reported by the probands to possess AP. Twenty-three siblings of respondents without self-re-

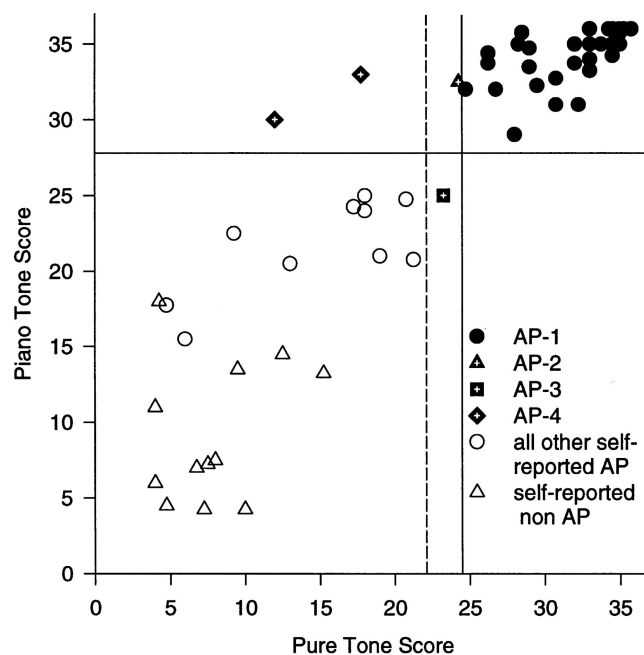
ported AP had received musical training at age <6 years, and only 2 were reported by the probands to possess AP. This difference between the AP status of siblings of AP possessors and that of siblings of non-AP possessors was significant ( $P = .0001$ ). These data support the conclusion that there may be a genetic component to AP.

### Testing for AP

To develop a quantitative measure of AP ability, we tested 48 self-reported AP possessors, using auditory tests designed in our laboratory. All the AP subjects tested claimed that they were able to accurately identify the pitch of tones in  $\leq 3$  s. The scores of these 48 individuals are shown in figure 2 (34 were designated as having AP-1, 1 as having AP-2, 1 as having AP-3, 2 as having AP-4, and 10 did not qualify for any of these categories). All the individuals who scored well on the pure-tone tests also scored well on the piano-tone tests; for example, all the AP-1 possessors scored  $>3$  SE above the mean for the piano-tone tests as well as the pure-tone tests. In contrast, some of the individuals who scored well on the piano-tone tests scored relatively poorly on the pure-tone tests (fig. 2).

Subsequent to the establishment of the above-described categories for AP, we tested an additional 51 self-reported AP possessors. Of these individuals, 35 were designated as having AP-1, 4 as having AP-2, 2 as having AP-3, 3 as having AP-4, and 7 did not qualify for any of the four categories of AP. The distribution of these 51 individuals, across the possible categories, was very similar to that observed in the first set of 48 self-reported AP possessors who were tested. When the entire sample of self-reported AP possessors was considered,  $\sim 70\%$  (69 of 99) were classified as having AP-1.

Interviews were conducted with the 99 AP probands who had taken the auditory tests. These interviews were used to determine the following information: their family pedigree structure, their ethnic background, and the history of musicianship among their family members. All probands were questioned regarding the AP status of other family members. In 11 families, at least one additional relative was readily available for testing and was tested (fig. 3). In 10 of the families in which a proband and another family member were tested, the individuals' scores placed them in the same group (all AP-1), including one family with five AP members (family 11) and another with four AP members (family 9). In one family (family 10), the score of the proband placed him in the AP-1 group, whereas his brother was placed in the AP-2 group. In five of these families, the probands reported AP in other family members whom we have not yet tested.

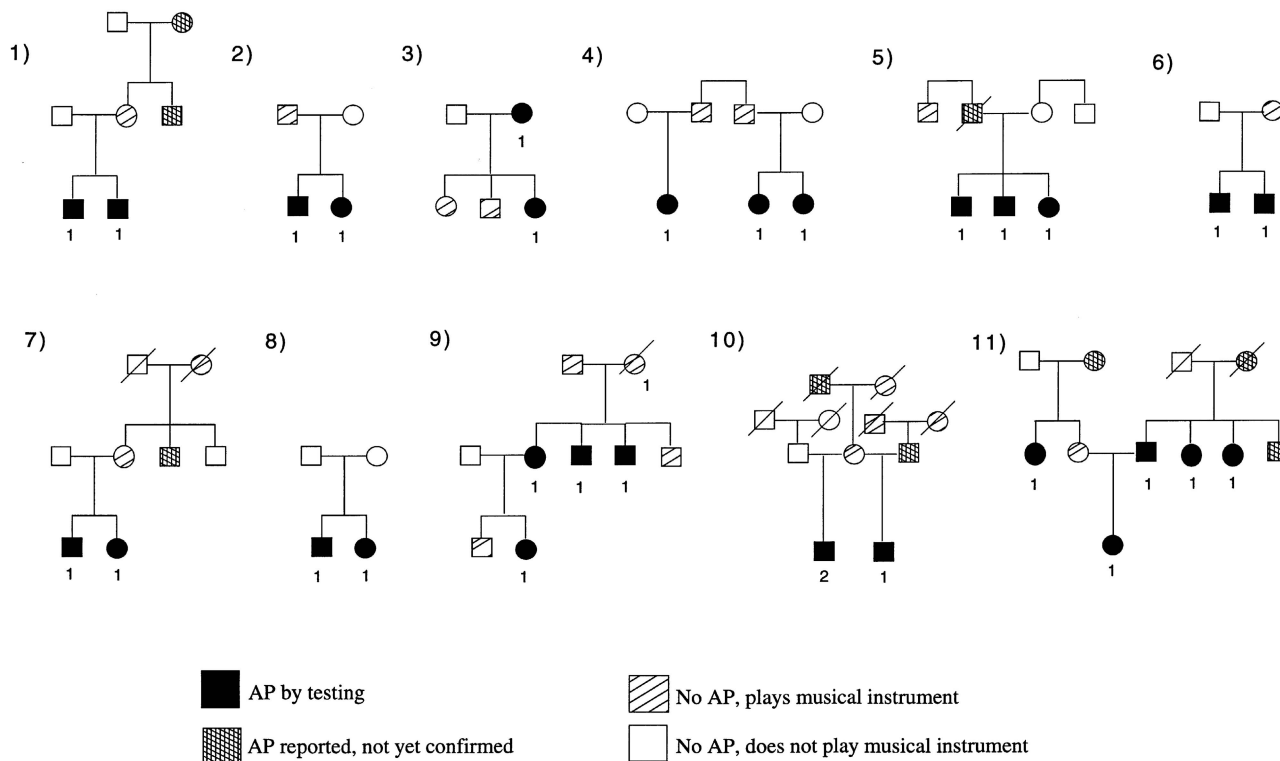


**Figure 2** Scatter plot produced on the basis of pure-tone and piano-tone scores of a combined sample of self-reported AP possessors and self-reported non-AP possessors, examined with auditory tests for AP. The maximum score obtainable for piano tones and pure tones was 36. Subjects' AP status was assigned as described in Subjects and Methods. The vertical dashed line indicates the mean pure-tone score  $+2$  SE; the vertical solid line indicates the mean pure-tone score  $+3$  SE; and the horizontal solid line indicates the mean piano-tone score  $+3$  SE.

### Discussion

This study was undertaken to characterize the AP phenotype, to set the stage for future studies aimed at identification of genes that predispose to AP. Our study has permitted us to draw broad conclusions about the development of AP, with more confidence than was possible in previous studies that were conducted with samples of only a few individuals with AP or with less rigorous criteria, and suggests that both inherited and environmental influences affect the development of AP.

Since some of our points are based on survey results, there are two caveats to our conclusions. First, although we attempted to sample in an unbiased manner, this process may not have been completely random; it is possible that response rates differed between individuals with self-reported AP and those without self-reported AP. Second, the survey subjects were asked to judge their own AP ability and that of their family members. Although we attempted to make the responses objective by asking specific detailed questions, some respondents may have been more critical than others in judging their own or their family members' AP ability. However, the



**Figure 3** Pedigree drawings for families of individuals with AP. For each pedigree, all available self-reported AP possessors were tested. Individuals were assigned to an AP category on the basis of their scores on our auditory tests, and the AP-group number is indicated below the symbol for each individual who was tested. Musical ability and AP status of the family members who were not available for testing were reported by the proband, for each family.

results of our auditory testing suggest that self-reports of AP were consistent with objectively measured AP ability, in most cases.

Previous studies have suggested that early musical training is the single most important factor for the development of AP (Takeuchi and Hulse 1993). Our study confirmed the importance of early musical training for the development of AP, since nearly all self-reported AP possessors stated that their formal musical training had begun at age  $\leq 6$  years. The correlation between early musical training and AP could be explained by a developmental critical period for AP, during which the brains of some individuals are particularly amenable to the establishment of new circuits or to the fine-tuning of pre-existing circuits involved in pitch perception. The existence of such a critical period has been demonstrated for singing behavior in songbirds and for language development in humans (Doupe 1993; Neville 1991). Alternatively, it is possible that individuals who are genetically predisposed to develop AP may be more likely than others to start musical training early in life. Thus, AP may be part of the general phenomenon of musicality, and an early interest in music could result from greater tonal acuity and increased awareness of sounds,

in predisposed children. Early musical training does not entirely explain the development of AP, since the majority of the respondents in our study who reported formal musical training at age  $\leq 6$  years also stated that they do not possess AP. Thus, our results are consistent with the hypothesis that early musical training is necessary but not sufficient for the development of AP.

The fact that most individuals with early musical training did not develop AP suggests a genetic contribution to the development of this phenotype. This possibility is supported by our observation that AP aggregates in families. Furthermore, in families for which two or more first-degree relatives with self-reported AP were tested, a strong concordance of the AP phenotype was noted. Similarly, other studies have suggested a genetic basis for AP (Profita and Bidder 1988; Gregersen and Kumar 1996). In the families that we have studied to date, the inheritance pattern of AP ability is compatible with the model suggested by Profita and Bidder (1988)—namely, autosomal dominant transmission with incomplete penetrance. Under such a model, the penetrance of AP may be influenced by early musical training.

The results from this study should provide the basis for the future identification of genes underlying AP. As

with any complex trait, mapping studies of AP will depend on the identification of a reliable phenotype that is likely to result from alleles in a small number of genes (McInnes and Freimer 1995). A stringent definition of AP, which identifies individuals with the fastest and most accurate pitch judgment, may provide us with one such phenotype. To that end, we have developed tests for AP that enable us to detect individuals with clear-cut AP (AP-1), on the basis of their highly accurate perception of both piano and pure tones. This category includes the majority of all individuals who self-report AP. Other self-reporting AP individuals (AP-2 and AP-3) have less reliable pitch perception. The individuals designated as having AP-4 appear to possess a distinct form of AP, compared with those in the other three categories; these individuals perform rather poorly on the pure-tone test but with almost complete accuracy in judging piano tones. The variability observed in the AP phenotypes may be related to the underlying processes involved in pitch perception in these individuals. For example, whereas AP-1 subjects can identify the pitch of tones simply on the basis of the fundamental frequency of the tones, AP-4 individuals may have an inherent ability to make use of the information provided by qualities such as the timbre and harmonics of tones, to accurately identify pitches. We propose to restrict future genetic mapping studies to AP-1 individuals and their families.

The understanding of any complex behavior requires dissection of its genetic and nongenetic elements. Methods are now available for elucidation of the genetic basis of complex traits (Lander and Schork 1994); however, the nongenetic factors that contribute to particular behaviors are often less well defined and hence more difficult to investigate. Therefore, human behavior is likely to be understood best by the study of traits for which it is possible to evaluate quantitatively both the genetic and the nongenetic factors that promote their development. In this paper, we provide evidence that AP is a model trait for the investigation of both the nature and the nurture of human behavior; we have confirmed that early musical training is essential for the development of AP and have operationally defined AP in a way that should allow us to identify genetic variations that can lead to the development of this fascinating ability.

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