Congenital Amusia: understanding of the musical mind and brain

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Abstract

Congenital amusia, commonly known as tone-deaf, is a developmental disorder that affects small population. The current paper aims to provide a comprehensive review of some of the recent researches on congenital amusia, demonstrating how the behavioural, brain-imaging, and genetic studies have extended our understanding of the musical mind and brain; additionally, identifies the gaps in the literature and suggests future research directions. Previous behavioural experiments and neurological studies argued that the deficit is congenital and arise from perceptual impairment in hearing fine-grained pitch, while genetic evidence lacked to pin point specific genes that are involved. Nevertheless, new studies show that the deficit may be improved through training, suggesting amusia may not be congenital after all.

Experience of music in every-day life comes effortlessly for most people. However, investigations over a decade suggests that a small population group are reported to be affected by a developmental disorder termed ‘congenital amusia’. The term implies that the deficit arises from birth and applies across broad impairment that links to musical perception as population of known individuals with amusia show large heterogeneous and convergent in the data (Peretz, 2016).

Congenital amusia manifests being unable to sing in tune to music, recognise mistuned notes in a melody, synchronise to a rhythm, or recognise familiar songs without lyrics (Ayotte, Peretz, & Hyde, 2002). Since the so called amusics show normal perceptual abilities, the musical disorder is independent to other auditory deficits such as hearing loss. The disorder is also irrelevant to lack of musical exposure or general learning ability as amusics acquire normal intelligence and memory, of them who often have had musical training in the past and formal lessons at schools (Ayotte et al., 2002).

Investigating the causal link of the deficits of amusics is like reverse-engineering. An impairment observed at a perceptual level can be traced back to the cognitive process and neurophysiological processes, which eventually leads to genetic level. For this reason, studying the developmental cognitive deficit in music sheds a light on the domain specific nature of music and extends to reveal on the shared neural pathways with other domains such as language.

This review will discuss some of the remarkable findings on the characteristics of impairment in congenital amusia – namely the perception of fine-grained pitch, contour (pitch direction), and musical memory. The discussion will extend to understand whether the deficit is specific to music or is a result of general impairment in psychoacoustics. Furthermore, drawing from recent brain-imaging and genetic studies, it aims to provide an overview of how investigating the deficit extended our understanding of the musical mind and brain. Finally, it will conclude by discussing some of the components that seem to be a gap in the literature and propose on future research directions.

Diagnosis: discriminating amusics

When investigating amusics, one of the advantages is that the recruitment of participants can be relatively easy. Usually the recruitment process is made through broadcasting on newspapers and online articles to target potential participants who believe that they acquire difficulty in experiencing music (e.g. Stewart, 2006). However, individuals feeling that they are ‘tone-deaf’ because they are unable to sing in tune to a song can be misleading. The disorder does not arise from insufficient ability of a single skill but usually from a

combination of deficit in music perception; however, a recent case study report suggest that the deficit can arise from a specific perceptual impairment such as rhythm, termed ‘beat-deaf’ (See Phillips-Silver et al., 2011). Because of this ambiguity in discriminating individuals with amusia, standardised criteria is necessary.

The Montreal Battery of Evaluation of Amusia (MBEA) (Peretz, Champod, & Hyde, 2003) is widely used as diagnosis for amusia. The battery is composed of famous tunes (without lyrics) lasting four bars in length with manipulated versions of these to test six distinct components in music perception. In this test, participants are asked to discriminate between the original and the systematically varied version of the tune, and each subset scores are summed to provide an individual global score.

In the first run of a group test on congenital amusia, almost 90% of self-reported individuals who considered themselves to have difficulty with musical experience scored below two standard deviations from the mean of matched comparison control group of 160 adults. Subsequent test and re-test of MBEA showed that the scores are normally distributed, and following studies adopting the test as the diagnosis confirmed that it is a reliable measure (Peretz et al., 2003).

Out of the six components in the test, three of them are related to pitch - namely contour, interval, and scale. This is consistent with a smaller study with 11 amusics that also reported deficits in all of the three components (Ayotte et al., 2002). The findings are no surprise as pitch is one of the fundamental aspects of music, and music exists across all known cultures (McDermott & Hauser, 2005). Thus, how may amusics perceive pitch differently to the normal population has been the focus of interest among many researchers.

Pitch is the Key

Fine-grained pitch discrimination

In Western music, consecutive notes in melodies can vary as little as one semi-tone and poor perception of this can affect the way one internalises a musical scale when listening to music. A single case study conducted by Peretz and colleagues (2002) of a master student named Monica, who reported to have difficulties in experiencing music, showed that she was unable to discriminate between two tones if they were less than two semitones apart. Moreover, Monica could not detect if the pitch was moving up or down – known as ‘contour’ in music.

Subsequently, Hyde and Peretz (2004) conducted a study with MBEA diagnosed 8 amusics and matched pairs of normal controls to examine the general threshold-level in pitch discrimination. Consistent with Monica study (Peretz et al., 2002), they reported that amusics could not discriminate pitch intervals below two semitones, whereas there was a ceiling effect

for controls around 0.25 semitones. In contrast, amusics performed as well as controls when the timing of the notes were varied (deviation from synchrony), indicating that the impairment is solely subjected to pitch information.

Follow up study using electroencephalographic method with event-related potential (ERP) analysis demonstrated that the pitch impairment can be traced down to electrical activity in the brain, and it provided deeper insight into the cognitive process in a neuronal level (Peretz, Brattico, & Tervaniemi, 2005). In this study, amusics performed the same pitch change discrimination task as Hyde et al. (2004) while taking measure of ERPs using electrode array. When the pitch change was small, amusic brain exhibited abnormal P3b ERP, component that is associated with discrimination of an attending target in a repeating sequence (Moreau, Jolicœur, & Peretz, 2013). Peretz, Brattico, Järvenpää, and Tervaniemi (2009) conducted a similar ERP study using anomaly tone detection task and observed abnormal P3b response to pitch deviance but normal mismatch negativity (MMN) exhibition, suggesting that their preconscious processing of change does not defer from normal individuals. Taken together, these studies suggest that amusic brain can track the small changes but lack in the conscious access to this change. This implies that the causal defect in pitch discrimination may lie outside the boundaries of auditory cortex region but extend to the auditory pathway.

Number of brain-imaging studies are converging to reveal that amusic brain exhibits abnormal activity in the right frontotemporal network, which involves the auditory cortex and the inferior frontal gyrus (IFG). Reduced connectivity has been observed between the auditory cortex and the right IFG (Albouy et al., 2013). Anatomically, higher density of grey matter and lower density of white matter has been observed in right IFG of amusics, as well as abnormal density of grey matter in the auditory cortex (Albouy et al., 2013; Hyde et al., 2007). Disturbance and altered transmission between these two regions of the brain and the left auditory cortex is regarded to be the cause of congenital amusia (Peretz, 2016). To sum, behavioural and cognitive neuroscience research highlight that the main deficit amusics experience is the fine-grained discrimination of pitch and reflects that the deficit is not domain specific to music, but rather more general psychoacoustic impairment.

Pitch discrimination in speech

Music and speech are similar in that both acquire temporal and pitch variations, where music is more largely tonal based (for overview on comparing acoustic cues for music and speech, see Zatorre, Belin, & Penhune, 2002). Hence, one may associate congenital amusia with a developmental disorder that exists in language such as dyslexia. In fact, there has been reports of language specific impairment that arise from difficulty in hearing fine temporal

variation in speech (i.e. Goswami, 2011). From this prospect, what temporal cues in speech may be is pitch to music. Nevertheless, in speech, pitch is modulated to convey different meaning (e.g. word stress, and focus) and rise in contour in a questioning statement also involves intonation in pitch (Eady & Cooper, 1986). Considering this, deficit of pitch discrimination in music may also extend to deficit in prosodic and utterance recognition in speech.

Ayotte et al., (2002) was the first to address this question and examined whether amusics are able to discriminate salient pitch accents in speech and statement from question. They demonstrated that amusics performed as well as controls in recognising speech but not music, concluding that the impairment is music specific. However, paradoxically, Patel and colleagues (2008) directed a similar study but reported a varied outcome, where 30% of British and French amusics could not discriminate the statements from questions. This raises even more question as 30% of amusics diagnosed through MBEA failed to discriminate, but the rest 70% performed as well as controls. Could it be that there exist various forms of the disorder, where one has domain specific deficit but the other extends also to language?

Liu, Patel, Fourcin and Stewart (2010) attempted to tackle this paradox by conducting a study incorporating statement and question items with relatively small yet ecologically valid pitch contrast. While controls performed well in detecting the subtle changes in the utterance and statement/question, all amusics performed poorly on both conditions. The finding indicates that amusics have no difficulty with speech in daily-life settings where the pitch intonations are generally large and sizeable enough that does not reach the pitch discrimination threshold; however, face difficulty in detecting subtle changes. Thus, varied performance reported by Patel et al. (2008) can be explained with differences in amusic individuals' ceiling threshold. A recent meta-analysis of 42 congenital amusia studies indicates that the contrast in pitch change is the largest determinant of discrimination-task performance for both music sequence and speech (Vuvan, Nunes-Silva, & Peretz, 2015). Taken together, in parallel with findings that specifically examined music, findings with correlates to speech implies that congenital amusia is not domain-specific but domain-general.

Outstanding question: what about tonal language speakers?

Most of the study described so far focused on Western population who speaks non-tonal language. However, for other tonal-languages such as Mandarin and Vietnamese draw heavily on pitch change as meanings in word differ according to the variation in contour. A study conducted with native Mandarin speaking amusics showed that they have no difficulty in discriminating words (Jiang, Hamm, Lim, Kirk, & Yang, 2011), but this is likely due to large

contrasts. Nevertheless, a recent study investigated tone-language speaking participants using fMRI and observed that, while controls exhibit significant activation in the right superior temporal gyrus (STG), no activation was detected for amusic (Zhang, Peng, Shao, & Wang, 2017). This reflects that acquiring tonal language may develop the neural structure associated with auditory pathway differently. As a result, neural deficit of tonal language amusics might differ from those non-tonal language speakers.

On a related note, native Mandarin and Vietnamese speakers are shown to have higher percentage of absolute pitch possessors in the population, presumably because of the tonal structure of their language (Schellenberg & Trehub, 2008). Statistical data estimates that congenital amusia affects about 4% of the people in the global population; however, newer estimate with much larger sample size reports around 1.4% (Peretz & Vuvan, 2017). With regards to the observed structural difference of some brain regions and higher percentage of absolute pitch possessors in the tonal-language population, future study should investigate the prevalence of congenital amusia in the tonal-language population group for a deeper insight of the disorder and its link to genetic origins.

Musical Memory

Some indications from studies suggested that amusia is not simply a low-level disorder and is beyond fine-grained pitch discrimination. Gosselin and colleagues (2009) used a pitch comparison task with two tones 1650ms apart, in which the retention interval between the tones were either left as a silent gap or filled with irrelevant notes. Whereas controls performed well in both conditions with up to three note intervals, amusics performed at chance level with one note interval. Interestingly, amusics performed as well as the controls when there was a silent gap for two and three note intervals, but performed poorly when irrelevant notes were present - suggesting that amusics are more likely to be affected by intervening tones and lack in 'pitch memory' (Schellenberg & Trehub, 2003).

The finding is consistent with an earlier study conducted by Foxton, Dean, Gee, Peretz, and Griffiths (2004), who demonstrated that amusics' memory for four-notes sequence is poorer than controls when the intervals within the sequence exceeds individuals' perceptual threshold. Moreover, amusics showed normal memory in digit spans, which indicates that the poor performance in pitch memory is separate from verbal short-term memory. Tillmann et al. (2009) also reported that amusics have deficits in memory of tone sequences and their timbral features but not with similar word sequences.

Nevertheless, these studies (Gosselin et al., 2009; Foxton et al., 2004; Tillmann et al., 2009) had limitation in that the testing of individual's threshold was carried out with fixed

length sequences. Williamson et al. (2010) brought an alternative approach to the table by employing an adaptive-tracking method to focus on individuals' capacity of pitch and verbal memory. This method allowed to measure individuals' span scores by examining performance at multiple sequence lengths, and it was suited for the purpose as there is likely to be a heterogeneity in the amusic population. In align with Foxton et al. (2004) and Tillmann et al. (2009), short-term memory task revealed that amusics performed as well as controls in the spoken digits conditions, but significantly worse with tones. In the working memory task, only subgroup of four individuals were found to have deficit in working memory but no evidence suggested for the rest. This implies that the subgroup of four participants may have a different deficiency from the majority of those diagnosed with amusia. Taken together, there could exist two scenarios on the musical memory deficit for amusics: 1) they lack in storing long-term memory of the musical structure that would otherwise help them to recall the information and maintain the tracking of tone sequences; 2) they can store the musical regularities but lack in the conscious access to this information, potentially due to the interrupted connection between auditory and frontal cortices (Plakke & Romanski, 2014).

Genetic Correlates of Amusia

Compared to extensive studies in a cognitive domain such as language, significantly less research has been undergone to look at genetic basis of musicality. In language, genetic investigation underlying speech disorder identified some genes (i.e. *FOXP2*) that are involved in the cause (Deriziotis & Fisher, 2013). As such, investigating congenital amusia sheds a light to understand the neurobiological underpinning of the pathway for music and more genetic studies are beginning to emerge.

Peretz and her colleagues (2007) investigated 9 large families of amusic probands with 71 members with matched control of 10 families with 75 members. The study showed that the disorder is a defect in pitch discrimination but not timing in music, and it is heritable. While the prevalence of the disorder was only 3% among first-degree relatives of control families, 39% was present in the amusic families. In a twin study of 136 monozygotic and 148 dizygotic twin pairs, participants were asked to discriminate the wrong note in a well-known song melody, and genetic model-fitting showed that shared genes were greater determinant than shared environment with estimate of 70 to 80% heritability (Drayna, Manichaikul, de Lange, Snieder, & Spector, 2001).

Likewise, some studies argue that individuals' musicality in general are more dependent to genetic basis compared to number of practice hours in the context of musical achievement (Peretz, 2016). Even the motivation to commit more hours of practice seems to

be genetically influenced. The topic of whether musicality is innate or shaped through the environment is an on-going debate.

Nevertheless, recent studies have also demonstrated that amusia can be improved through laboratory training and raises a more interesting question (Liu, Jiang, Francart, Chan, & Wong, 2017; Whiteford & Oxenham, 2017, 2018). In a study conducted by Whiteford and Oxenham (2017), 20 amusics and matched pair of controls undertook four sessions to train in pitch-discrimination task. After the training, 11 of the amusics no longer met the criteria of MBEA and one year follow up test showed that the improvement is maintained (Whiteford & Oxenham, 2018). This is controversial to the previous findings that amusia is a life-long deficit and questions the current diagnosis of MBEA and whether the disorder is influenced by genetics.

Closing Remarks and Future Directions

The current review aimed to provide a comprehensive review of some of the recent researches on congenital amusia and to demonstrate how the behavioural, brain-imaging, and genetic studies have extended our understanding of the musical mind and brain. Both behavioural and neurological studies highlight that the disorder arise from deficit in fine-pitch discrimination and the cause of this is likely to be the disturbed transmission between the right IFG and the left auditory cortex. Moreover, comparison between speech and music with pitch contrast indicate that the impairment is not domain-specific but general psychoacoustic defect of frequency resolution. The disorder seems to go beyond the low level cognitive process as impairment in pitch memory has also been observed. Genetic studies demonstrate that the deficit is heritable, however, new findings are beginning to emerge that training can improve the disorder, suggesting that it may not be ‘congenital’ after all for some individuals.

There are few points that concerns to be a gap in the literature of congenital amusia that require attention. Firstly, although investigation in emotional aspect and ability to engage in music for amusics are beginning to emerge (Omigie, Müllensiefen, & Stewart, 2012) and survey studies have been conducted (McDonald & Stewart, 2008), hardly no qualitative research has appeared in the field. Future studies incorporating interviews studies with amusics may reveal novel questions and new pathways. Secondly, concerning fine-grain pitch discrimination, most studies focused on pitch intervals within the range of around two octaves that is centred around midline of the piano (200 ~ 600 Hz). Examining across the whole range of the frequency spectrum may reveal more specific characteristics of the deficit. Previous researches demonstrate that, for general population, discriminating tones become more difficult

in the extreme boundaries of the audible frequency range (20 ~ 20,000Hz) (Pressnitzer, Patterson, & Krumbholz, 2001). Whether the difficulty follow the same or different curve pattern for amusics and examination of this may able to pin point a specific frequency boundary that is more affected by the disorder.

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