Pitch Discrimination and Self-Reported Musicality in 22q11.2 Deletion Syndrome

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Background & Rationale
Chromosome 22q11.2 deletion syndrome (22q11DS) is a genetic disorder with varying phenotypic manifestations ranging from congenital cardiac, palatal, and skeletal anomalies to psychopathologies such as ADHD, anxiety, mood disorders, and schizophrenia. Observations from a longitudinal collaborative study at the University of Pennsylvania and CHOP suggest that for some participants with 22q11DS, participation in musical activities is common. While no previous studies have examined musicality in 22q11DS, deficits in pitch discrimination have been observed in those with schizophrenia, thus linking musicality with psychopathology. Our original aim was to analyze the musical abilities of 22q11DS participants compared to healthy controls; however, as assessments of healthy controls and data validation of recent scores have yet to be completed, the following analysis focuses solely on 22q11DS individuals and the link between their psychopathological diagnoses and musical abilities. We hypothesize that the presence of psychopathology is associated with impaired musical abilities.

Method
Thirty 22q11DS participants (mean age = 15.8±5.3 years) completed the Goldsmith Musical Sophistication Index (GMSI), a self-report questionnaire assessing five musical domains: Active Engagement, Perceptual Abilities, Musical Training, Singing Abilities and Emotional Response to music. They also completed the Distorted Tunes Test (DTT), a computerized test of 26 melodies that assesses pitch discrimination abilities. The accuracy (number of correct responses) and response time (median time for correct responses) from the DTT were recorded. Additionally, all participants were assessed for psychopathology using semi-structured interviews.

Analysis and Results
We examined the associations of presence/absence of psychopathology (ADHD, Anxiety Disorders, Mood Disorders, and Prodromal/Psychotic Symptoms) to GMSI scores using Fisher’s Exact Test and to DTT accuracy and response time using ANOVA. We found that ADHD was significantly associated with scores in Musical Training ($p=0.03$) such that presence of ADHD was associated with lower scores in Musical Training. The presence of ADHD was also significantly associated with lower scores in Perceptual Abilities ($p = 0.01$). There were no other significant associations among other psychopathologies, GMSI and DTT measures.

Conclusion
A larger sample size is needed to conclude on the clinical significance of these results. Assessments are still ongoing. The inclusion of the control group will help to compare the musical abilities of 22q11DS individuals to healthy individuals, thus giving a more complete picture of musicality in this population.