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An fMRI study of facial emotion processing in children and adolescents with 22q11.2 deletion syndrome

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Introduction: 22q11.2 deletion syndrome (22q11DS) is a genetic disorder associated with interstitial deletions of chromosome 22q11.2. In addition to high rates of neuropsychiatric disorders children with 22q11DS have impairments of face processing, as well as IQ-independent deficits in visuoperceptual function and social and abstract reasoning. These face processing deficits may contribute to the social impairments of 22q11DS. However, their neurobiological basis is poorly understood.

Method: We examined 14 children and adolescent with 22q11DS and 14 healthy controls, using event-related fMRI to examine neural responses when they incidentally processed neutral expressions and mild (50%) and intense (100%) expressions of fear and disgust.

Results: Both groups significantly activated 'face perception' areas when viewing neutral faces, including fusiform-extrastriate cortices. Further, both groups had significantly increased activation of extrastriate-fusiform cortices and cerebellum to increasing intensities of fear. To increasing intensities of disgust, however, children with 22q11DS showed increased activation of fusiform-extrastriate cortices, whereas the controls had significantly decreased activation of cerebellum and other cortical regions. Moreover, children with 22q11DS generally showed reduced activity in brain regions involved in social cognition and emotion processing compared to controls across emotion types and intensities.

Discussion: Regions involved in face processing, including fusiform-extrastriate cortices, anterior cingulate gyri, and supramedial prefrontal cortices (BA 6), are activated by facial expressions of fearful, disgusted, and neutral expressions in children with 22q11DS, but generally to a lesser degree than controls. Activity in some of these regions also negatively correlated with extent of social difficulties. Hypoactivation in these regions may partly explain the social impairments of children with 22q11DS.

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Electrophysiological similarities in Tourette syndrome and chronic tic disorder

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Introduction: Tourette syndrome (TS) is a neuropsychiatric disorder characterized by both multiple motor tics and at least one phonic tic. Chronic tic disorder (CTD) is characterized by either motor or vocal tics, but not both. These two conditions are part of the “tic disorders” category in the DSM-5. However, the distinction between these two conditions is only based on tic symptoms, and not on a neurophysiological basis. The need for a distinction has been debated, since phonic tics have an inherent motor component. Also, the “wax and wane” nature of tics is another limit to the diagnostic. It is possible for one to face both motor and phonic tics at one time, and to only have motor tics at another moment of his life. To our knowledge, only one study has compared these two groups. While using neuropsychological measures, they found no difference between TS and CTD patients.

Aims: The present project aims to evaluate the event-related potentials in TS and CTD patients, to find similarities or differences in cerebral activation between the two groups.

Method: Cerebral activation was assessed in 3 groups (TS, CTD and control) using event-related potentials (ERP) during a stimulus-response compatibility task.

Results: There was a high cortical activation that was similar in TS and CTD patients, and corresponded to the response generation. This activation was mostly in central and parietal areas. This pattern of activation was not observed in control participants.

Conclusion: These results suggest that there is no neurophysiological difference between TS and CTD. The stimulus-response incompatibility affects both groups in the same way, while control participants present a lower degree of activation. TS and CTD patients might maintain a high level of motor activation to process incompatible stimuli while generating their response.

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