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A comparative study of the neuropsychiatric and neurocognitive phenotype in two microdeletion syndromes: velocardiofacial (22q11.2 deletion) and Williams (7q11.23 deletion) syndromes.

Zarchi O[1], Diamond A[2], Weinberger R[3], Abbott D[2], Carmel M[4], Frisch A[4], Michaelovsky E[5], Gruber R[6], Green T[7], Weizman A[8], Gothelf D[9].

Abstract

PURPOSE:

22q11.2 deletion syndrome (22q11.2DS) and Williams syndrome (WS) are common neurogenetic microdeletion syndromes. The aim of the present study was to compare the neuropsychiatric and neurocognitive phenotypes of 22q11.2DS and WS.

METHODS:

Forty-five individuals with 22q11.2DS, 24 with WS, 22 with idiopathic developmental disability (DD) and 22 typically developing (TD) controls were compared for the rates of psychiatric disorders as well as cognitive executive and visuospatial functions.

RESULTS:

We found that while anxiety, mood and disruptive disorders had an equally high prevalence among individuals with 22q11.2DS, WS and DDs, the 22q11.2DS group had the highest rates of psychotic disorders and the WS group had the highest rates of specific phobia. We also found that the WS group demonstrated more severe impairments in both executive and visuospatial functions than the other groups. WS and 22q11.2DS subjects had worse Performance-IQ than Verbal-IQ, a feature typical of non-verbal learning disorders.

CONCLUSION:

These findings offer a wide perspective on unique versus common phenotypes in 22q11.2DS and WS.

KEYWORDS:

22q11.2 deletion syndrome; Executive functions; Psychiatric manifestation; Visuospatial functions; Williams syndrome