22q11 Deletion syndrome and neurotransmitter systems in unchallenged and challenged conditions
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CHAPTER 8

Co-occurrence of early-onset Parkinson's disease and 22q11 deletion syndrome

Submitted

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Co-occurrence of early-onset Parkinson’s disease and 22q11 deletion syndrome

Catechol-O-methyltransferase (COMT) is responsible for dopamine clearance. Adults with 22q11 deletion syndrome (22q11DS) are hemizygous for the COMT gene, and consequently show increased dopamine levels [3]. Contrarily, Parkinson’s disease (PD) is characterized by dopamine loss [2]. Recently, co-occurrence of early-onset PD in two unrelated 22q11DS individuals was reported [4]. This was unexpected given the presumed opposite dopamine levels, and both conditions are relatively uncommon (for prevalences see [4]). Although a chance association between both conditions remains possible, an etiologic association was suggested [4]. Consequently, identification of further cases is helpful to study a possible association. However, adults with 22q11DS are frequently treated with neuroleptics which may hinder a reliable clinical PD diagnosis. Here we present another case of co-occurrence of early-onset PD and 22q11DS, in which dopamine transporter (DAT) imaging was helpful to support the PD diagnosis.

This 52-year-old male has a longstanding history of learning disabilities and psychotic disorder. Fluorescence in-situ hybridization demonstrated a 22q11 deletion. Additionally, over 10 years he suffered from parkinsonism, which was considered a side effect of neuroleptic treatment or a clinical feature of early-onset PD. However, the patient was not able to quit neuroleptics which hindered the clinical diagnostic process. $^{[123]}$I-FP-CIT SPECT showed striatal DATs loss (Figure; transversal slices; left-panel: 55-year-old male control; right-panel: 22q11DS case), supporting co-occurrence of early-onset PD in this 22q11DS patient.

We show that DAT imaging helps to differentiate neuroleptic-induced parkinsonism from PD [1], and can be used to study a possible etiologic association between 22q11DS and early-onset PD.
REFERENCES


