22q11.2 deletion syndrome diagnosed in a young patient presenting with Parkinson disease

Chromosome 22q11.2 deletion syndrome (22q11.2DS), also called DiGeorge syndrome, is a fairly common genetic syndrome. In most cases, the diagnosis is made at birth or early in childhood as the most common features of the syndrome – heart abnormalities, facial and palatal defects, developmental delay, and frequent infections – usually appear at an early age. The symptoms associated with 22q11.2DS, however, are not only seen in childhood; patients with this syndrome often develop other medical conditions when they reach adulthood. The most common adult-onset conditions in these patients include schizophrenia, bipolar disorder, seizures, and hormone and reproductive issues.

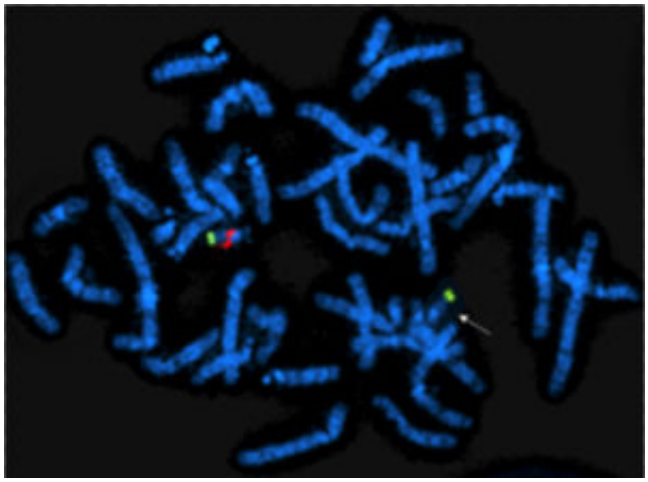

Fig. 1. A chromosome spread with a green marker identifying two copies of chromosome 22. One chromosome is normal, containing the critical 22q11 region in red; the other chromosome is abnormal (arrow), missing the critical red region.

Recently there have been reports suggesting that early-onset Parkinson disease (PD) may be yet another medical condition that 22q11.2DS patients can develop in adulthood. PD is a neurologic movement disorder associated with tremors, slow movement, muscle stiffness, and problems with balance and walking. It is most commonly a disease of the elderly, affecting people over the age of 60. In less than 5% of patients, however, symptoms develop before the age of 40. These cases are called early-onset PD.

We encountered one such case of early-onset PD in a young woman who developed tremors at the age of 29 and was eventually diagnosed with PD at the age of 34. Although otherwise healthy, her past medical history was notable for two childhood conditions commonly associated with 22q11.2DS: premature fusion of her skull bones requiring surgical repair at the age of 4 months,

and a heart defect referred to as the Tetralogy of Fallot requiring surgical repair at the age of 3 years. Except for some mild developmental delay, her childhood and development were otherwise normal and she was never diagnosed with 22q11.2DS.

Given her childhood conditions and presentation with early-onset PD, she was referred for genetic testing. A chromosomal analysis indeed revealed a deleted portion of chromosome 22 consistent with 22q11.2DS, as seen in Figure 1.

Most case reports to date of 22q11.2DS patients who have early-onset PD describe patients with known 22q11.2DS who later are diagnosed with PD, often in the setting of treating psychiatric disorders with medications that can cause or worsen the symptoms of PD. Our case was unique because our patient first presented with early-onset PD, which then led to a diagnosis of 22q11.2DS at age 34 rather than in early childhood.

A diagnosis of 22q11.2DS is important to make as early as possible because the disorder can lead to a variety of chronic illnesses that require ongoing medical care and because some medications for psychiatric disorders may need to be avoided. Our patient's story adds to growing evidence of a link between 22q11.2DS and early-onset PD and that screening for this genetic syndrome should be considered in patients who present with PD at a young age.

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