p-178

Saturday

Klinefelter syndrome: a novel cause of the behavioral variant of frontotemporal dementia phenocopy syndrome

Sara Rubio Guerra, Javier Arranz, Nuole Zhu, Mª Belén Sánchez-Saudinós, Isabel Sala, Javier Pagonabarraga, Íñigo Ruiz Barrio, David Almenta, Josefina Pérez Blanco, Sara Bernal Noguera, Oriol Dols Icardo, Valle Camacho, Juan Fortea, Daniel Alcolea, Alberto Lleó, Ignacio Illán Gala

The behavioral variant of frontotemporal dementia (bvFTD) is a neurodegenerative syndrome characterized by progressive behavioral impairment and functional decline. The bvFTD phenocopy syndrome refers to a subgroup of patients (mostly men) of uncertain etiology due to a lack of neuroimaging changes and clinical deterioration.

We present a case report of an adult male with the bvFTD phenocopy syndrome who was subsequently found to harbor a 47,XXY karyotype. A literature review was conducted on the cognitive and behavioral changes in adults with Klinefelter syndrome (KS).

He presented at the age of 65 with a one-year history of personality change comprising apathy, loss of empathy, resumed smoking, and new-onset gambling. He had partially preserved insight and performed independently in activities of daily living at baseline and on follow-up. He was sterile and had no first-degree family history of neuropsychiatric conditions. Neuropsychological assessment revealed prominent executive dysfunction. Neurological examination was unremarkable except for a mild postural tremor. No atrophy/hypometabolism was detected in neuroimaging studies. Alzheimer's disease pathophysiology and major FTLD-causing genes were ruled out. The observation of postural tremor lead to targeted *FMR1* testing that excluded Fragile-X syndrome but pointed at the duplicity of the X chromosome, and KS was subsequently confirmed. KS is largely underdiagnosed and classically presents with hypergonadotropic hypogonadism, testosterone deficiency, and infertility, but impairment in behavior, social cognition, and executive function are also recognized.

KS must be suspected in sterile males with the bvFTD phenocopy syndrome. Future studies are warranted to examine the frequency of KS in these cases.

Conflicts of interest

N/A