

CORRESPONDENCE

Klinefelter Syndrome: The Commonest Form of Hypogonadism, but Often Overlooked or Untreated

by Prof. Dr. med. Dr. h.c. Eberhard Nieschlag in volume 20/2013

Psychological and Neurological Problems

Although all of the somatic comorbidities reported in the article (1) occurred at rates below 50%, “speech impairment”, “dyslexia,” and “learning difficulties” reached rates of 50–80%. Furthermore, according to Bruining et al (2), ADHD is found in 63%. That means that the absence of such disorders is the exception rather than the rule, so that it’s possible these are not comorbidities but, rather, symptoms of the entity “Klinefelter syndrome” (KS). According to Savic (3), KS can serve as a model for androgenic effects on the development and function of the human brain. The behavioral phenotype is characterized by speech, executive, and psychomotor dysfunction and socio-emotional impairment. Neuroimaging studies in children and adults with KS have shown characteristic structural differences compared with normotypical individuals. In addition to an increase in the volumes of gray matter in the sensorimotor and parieto-occipital regions, significant decreases have been observed in the region of the amygdala and the hippocampal, insular, temporal, and frontobasal regions. AN increase in white matter is seen in the left parietal lobe, a decrease, among others, bilaterally in the area of the anterior cingulum. For the adequate treatment of KS, testosterone substitution is not the only therapeutic modality, but diagnostic evaluation and treatment by a child and adolescent psychiatrist and psychotherapist are essential, especially since 32% of KS patients have anxiety disorders, 27% autism spectrum disorders, and 24% depressive disorders (2). Van Rijn et al (4) found clear indications of autism spectrum disorders in 48% of adults with KS; consequently, adults with KS similarly require an examination by a specialist for psychiatric and psychological disorders who is familiar with autism disorders.

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Dr. med. Ingo Spitzcok von Brisinski

Psychiatrie, Psychosomatik und Psychotherapie des Kindes- und Jugendalters LVR-Klinik Viersen, Ingo.SpitzcokvonBrisinski@lvr.de

Conflict of interest statement

Dr Ingo Spitzcok von Brisinski has received honoraria for speaking about psychiatric disorders from ADHS Deutschland [ADHD Germany], Autismus-Therapiezentrum [center for autism therapy] Münster, Kompetenznetzwerk Autismus Oberschwaben [competence network for autism, Upper Swabia], and Janssen-Cilag.

Cardiological Disorders

The review article highlights many clinical aspects (1). However I missed aspects of cardiological disorders, especially in differential diagnostic distinctions from other neurological disorders. I had opportunity to observe a case of Wolff-Parkinson-White syndrome in a young patient with KS, in whom attacks of tachycardia resulted in impaired consciousness. Apparently, a statistically increasingly frequent prevalence of WPW syndrome (and perhaps other heart arrhythmias) has been observed for Klinefelter syndrome.

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Dr. med. Reinhard Niemann

Praxis für Neurologie und Psychiatrie, Kinder- und Jugendpsychiatrie sowie Betriebsmedizin, Radolfzell

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The author declares that no conflict of interest exists.

In Reply:

As explained in our article, Klinefelter syndrome (KS) is characterized by numerous comorbidities. These include cardiac disorders, which thus far have been seen mostly in association with the metabolic syndrome. The coincidence of KS and Wolff-Parkinson-White (WPW) syndrome, as described by Dr Niemann, seems to be a very rare event because WPW syndrome was not found in any of the 69 Italian patients with KS who underwent intensive cardiological investigations (1), nor was it described in any of the more than 500 patients in our institution, and, notably, it was not found in any of the specially cardiological investigated subgroup of 132 patients. However, it should be pointed out that disorders of the heart and vasculature contribute to increased mortality in patients with KS.

Dr Spitzcok von Brisinski expands on the section on psychological and neurological problems in boys with KS before puberty; we kept this short in our article for reasons of space. The literature he cites also complements the citations in the review article very appropriately. The letter re-emphasizes that KS should also be considered in children and adolescents with psychological and psychiatric abnormalities and thus supports the intentions of our review article.

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Prof. Dr. med. Dr. h. c. Eberhard Nieschlag FRCP

Centrum für Reproduktionsmedizin und Andrologie, Universitätsklinikum Münster Eberhard.Nieschlag@ukmuenster.de

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