Mayer-Rokitansky-Küster-Hauser Syndrome and Psychiatric Conditions: Toward the Importance of a Genetic Link

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Letter to the Editor

Mayer-Rokitansky-Küster-Hauser Syndrome and Psychiatric Conditions: Toward the Importance of a Genetic Link

TO THE EDITOR: We recently read with interest the article by Heller-Boersma et al.¹ published in *Psychosomatics* and entitled “Psychological Distress in Women with Uterovaginal Agenesis”. This might be one of the very first studies that has formally investigated the relationship among Mayer-Rokitansky-Küster-Hauser Syndrome (MRKHS), psychological distress and/or psychiatric conditions. The authors concluded that women with MRKHS ($n = 66$) had significantly higher pathological scores on subscales of the Symptom Checklist-90-R9 scale, such as phobic anxiety ($P = 0.02$) and psychoticism ($P = 0.054$), compared with controls ($n = 31$). Using the Eating Disorder Inventory, it appeared that MRKHS women have a significantly greater risk to develop bulimia ($P = 0.017$), which could be explained by them trying to compensate with food for lowered self-esteem and interpersonal difficulties. It is assumed that the most critical period for MRKHS patients is right after a surgical operation or at the time of the diagnosis, when they are most vulnerable to psychiatric symptoms. Unfortunately, in this

study, psychiatric evaluation was carried out a long time after diagnosis (9.6 years), which is the reason why the authors explain the lack of more dramatic differences in comparison with the control group.

Thus, we were interested to highlight what has been done on the link between MRKHS and psychiatric disorders during the past 10 years by the scientific community, although, because of its low prevalence rate, only few studies have been accomplished in the field.

Laggari et al.² established significantly higher scores for MRKHS patients ($n = 24$) for anxiety symptoms (State-Trait Anxiety Inventory) compared with controls ($n = 24$). The authors explain their results by the diagnosis and loss of reproductive ability that, especially in late adolescence, undermines emotional stability, physical maturity and the end of sexual identity development.

Though rarely studied, the association with schizophrenia (and psychosis) is no less interesting. Indeed, only three case studies of this kind have been reported. In 2012, Melliou et al.³ showed an association between sexual delusion and MRKHS. They reported the case of a patient with a treatment-resistant paranoid schizophrenia. The onset age was 20 years, and the diagnosis was made three years after the vaginal surgery. Her main delusion was to have sexual intercourse with an eminent person through the big toe of her right foot. From that, two main hypotheses emerged. The neurological one, suggesting that the deactivation of the patient's genitalia

led to an expansion of the adjacent cortical area. In the somatotopic arrangement, the two territories are near each other. The psychodynamic hypothesis supports that the sexual function was expelled from the body image and was stored in a nonsexual part of the body.

In 2015, Bhad et al.⁴ published the second case of this kind but with a later age of onset (24 years) and before vaginoplasty. Beside delusion of control, impulsivity and suspiciousness, the diagnosis of schizophrenia was largely based on a sexual delusion (sensations in the vaginal area attributed to sexual intercourse with an unknown man living abroad and controlling her mind). Interestingly, they suggested a possible genetic link between these two diseases through a pathogenic copy number variation: the 17q12 deletion. Indeed, mutation in this area, at the DNA level, is associated with a greater risk of both schizophrenia and MRKHS.

Finally, Nath et al.⁵ presented in their case report a 22-years-old woman with diagnosis of unspecified nonorganic psychosis, diagnosed with MRKHS in the meantime, having a good response to olanzapine and divalproex sodium bitherapy. Besides acknowledging that stress (caused by the physical deformity) can lead to psychological disturbances, the authors suggested a genetic link between MRKHS and psychiatric disorders and suggested that a pathogenic copy number variation could, again, be considered as a double risk factor for both physical and psychological conditions.

Letter to the Editor

The above mentioned articles reinforce the original findings of the article published 10 years ago by Heller-Boersma et al.¹ However, there is something new in the diagnostic approach. Owing to progress in the field of genomics, with indications of shared genetic risk factors, it might be an interesting new perspective to draw more attention to the genetic background of the MRKHS patients. New research, designed to highlight evidence of these risk factors, could lead to identify MRKHS patients with high-risk profiles for psychiatric outcomes. Thereby, through these data, systematic earlier psychological evaluations (mostly before surgery) and support could improve the global quality of care.

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