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Mini-Review

Mayer-Rokitansky-Küster-Hauser Syndrome: Sexuality, Psychological Effects, and Quality of Life

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Abstract. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital condition in which a genetic female is born with vaginal agenesis and a rudimentary to absent uterus. This condition affects a woman's ability to menstruate, to engage in penile-vaginal intercourse, and to bear children. Much has been published about how best to create a neovagina in women with MRKH, but little has been written about the psychological impact of MRKH and quality of life outcomes for women with the condition.

A review of the extant literature published from 1955 to 2007 supports that (1) surgical or non-surgical creation of a neovagina alone does not ensure a successful psychological outcome, (2) psychological support at critical times can be helpful, and (3) how professionals use language to discuss the condition may positively or negatively influence a female's experience of MRKH. This article discusses the implications that existing knowledge has on future research and on clinical practice. Understanding how women with MRKH cope with and adjust to the condition will help healthcare professionals provide optimal care.

Key Words. Mayer-Rokitansky-Küster-Hauser syndrome—MRKH—Vaginal agenesis—DSD—Psychology—Sexuality—Quality of life

Introduction

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is an uncommon, but not rare, congenital anomaly of the female genital tract, estimated to occur in approximately 1 in every 5,000 females.[1] Features include vaginal agenesis and uterine abnormalities that range from an absent uterus to rudimentary uterine remnants. Affected females usually have functioning ovaries, normal external genitalia, and a female karyotype (46,XX). MRKH syndrome is the second most frequent cause of amenorrhea after gonadal dysgenesis[1] and is often discovered when a patient presents in adolescence due to primary amenorrhea.

MRKH syndrome is one of many Disorders of Sex Development (DSD). DSD refers to "congenital conditions in which the development of chromosomal, gonadal, or anatomic sex is atypical."[2] DSD is a relatively new term and was proposed by international experts from the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology at the consensus meeting in October, 2005. DSD replaces out-of-date nomenclature like intersex and hermaphrodite. This review was undertaken partially in response to a request in the Consensus Statement on the Management of Intersex Disorders, a summary of the consensus meeting, that more attention be focused on the psychological aspects of DSD.[2] MRKH syndrome, like other DSD, poses challenges that go far beyond physical concerns. Since the publication of the Consensus Statement much dialogue has been generated. Often noted is the lack of long-term outcome studies for those with DSD.[3] Additionally, it has been recommended that a shift in emphasis takes place from the physical aspects of DSD to how individuals adjust to the conditions.[3] This review supports this shift and the position that psychological issues as well as medical aspects must be addressed in order to provide optimal care.

A young woman's sense of well-being and quality of life are impacted by the condition. Affected individuals without treatment will find it difficult to engage in penile-vaginal intercourse, do not menstruate, and will be unable to carry a pregnancy. The discovery that sexual intercourse will not occur without medical intervention and the realization of loss of childbearing may be devastating to an adolescent who has not yet reached certain developmental milestones. Because of physical

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and psychological challenges, it has been suggested that the management of MRKH falls into two categories: (1) the need to anatomically manage the anomaly so that young women may have the option to engage more easily in penile-vaginal intercourse and (2) the need to help young women cope with the psychological impact of the condition.[4] Although the outcomes of surgical and non-surgical treatments to create a new vagina have been reported, there have been few reports addressing the psychological impact of the condition and quality of life (QOL) outcomes.

The aims of this article are three-fold: (1) to summarize the extant literature on the psychological effects of MRKH and QOL outcomes; (2) to identify existing gaps in the literature regarding the psychological effects of MRKH and QOL outcomes; and (3) to offer suggestions for future research and clinical practice for women with MRKH.

Methods

A search of the databases Medline, Psyc INFO, PsycLIT, and CINAH was conducted. The following keywords were used: Mayer-Rokitansky-Küster-Hauser syndrome, MRKH, vaginal agenesis, vaginal aplasia, psychological outcomes, gender identity, gender role, sexual orientation, sexual functioning, infertility, conception, marriage/cohabitation, social functioning, cognitive functioning, psychopathology, self-concept, career, spirituality, legal history, family response to MRKH, and individual adjustment to MRKH. From 36 articles collected, 31 were reviewed for this article.

Inclusion

Articles were included that provided data on the psychological and psychosexual effects of MRKH as well as any information about a young woman's quality of life after diagnosis and treatment of MRKH. Data in the studies had to pertain to a diagnosis of MRKH. Only English manuscripts and those translated into English published between 1955 and 2007 were included. The year of 1955 was selected as the starting point for this review because this was the year that John Money published his now classic work on hermaphroditism,[5,6] now referred to as DSD.

Exclusion

Data in which the results were not separated by specific diagnosis were excluded. For example, studies by Evans et al,[7] Hensle et al,[8] and Liao et al[9] were excluded because the data from those subjects who had MRKH could not be separated from the data of those participants who had other etiological reasons for vaginal malformations such as Androgen Insensitivity syndromes (AIS), Mixed Gonadal Dysgenesis, or cancer treatments.

Results

The 31 articles reviewed were divided into several categories. There were 11 articles that focused primarily on how the condition of MRKH affects young women psychologically. Another 18 articles reviewed specific treatments to create a neovagina, and included varying amounts of information about how young women adjust psychosexually, emotionally, and/or psychosocially to the condition. In addition, two articles addressed the ability to achieve pregnancy through surrogacy.

The Centre for Health Promotion (2007) at University of Toronto conceptualizes QOL as the ''degree to which a person enjoys important possibilities of his/her life''[10] taking into consideration a person's physical, psychological, social, and spiritual dimensions.[10] Having the condition of MRKH affects a woman's quality of life by placing limits on some of life's possibilities (like intercourse and childbearing) and may cause distress and an altered self-concept. For this article, attention was given to studies that reported on how women were functioning in regards to specific QOL domains. Also, there was a focus on how women were impacted psychologically by the limitations that MRKH imposes. The results are organized according to these dimensions of QOL and psychological effects.

Quality of Life Outcomes

Sexuality

Sexual functioning was addressed in 91% of the studies. The primary aim in most studies was to evaluate a specific treatment (either surgical or non-surgical) to create a neovagina. Because of the large number of articles that evaluated treatment techniques these data are reported in table format (see Table 1).[11–31] The focus of these articles ranged from the anatomical success of a neovagina to a more qualitative focus that included orgasm and natural lubrication. Additionally, by looking at the outcomes of these various treatments, data were also gathered about patients' psychosexual functioning and sometimes the emotional and psychosocial impact of the condition.

The literature suggests that after treatment, most women who underwent one of the many surgical or non-surgical techniques to create a neovagina were able to engage successfully in penile-vaginal intercourse; indicating that the vagina was long enough and wide enough to accommodate an erect penis. There were many reports of orgasm[12–15,17,22,26,28,29] and adequate natural lubrication[13,18,19,21–24,29] that indicated women with MRKH were actively participating in coitus. An affirmative response to the question, ''Are you sexually active?'' implied that