Multidisciplinary health care for women with Mayer-Rokitansky-Küster-Hauser syndrome

Since the publication of the Dutch guidelines on primary amenorrhea, we evaluated our 10-year experience with respect to the provision and utilization of multidisciplinary care for women* with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome in a center of expertise (1).

MATERIALS AND METHODS

A descriptive report by medical file review was performed. Women with MRKH syndrome were included if they had at least 1 gynecological consultation regarding the diagnosis or neovaginal treatment at Radboud University Medical Center (Radboudumc) between 2012 and 2021. The data on baseline characteristics, diagnostics, therapy, social embedding, psychological aspects, sexual aspects, multidisciplinary approach, and follow-up were extracted and analyzed.

RESULTS

A total of 112 files were screened for eligibility, and 85 medical files were included for analysis. Table 1 summarizes the characteristics, diagnostics, and treatment of women with MRKH syndrome who presented at Radboudumc between 2012 and 2021. The median age at diagnosis was 16 (range, 12–29 years) years, with primary amenorrhea as the main reason for the first consultation. In 72% (n = 59) of women, diagnosis was made in secondary care. Ninety-two percent (n = 71) underwent magnetic resonance imaging, the golden standard in diagnosing Müllerian anomalies, with a renal anomaly noted in 19% (n = 16) of our study population (2, 3). Referrals to a psychologist, sexologist, and pelvic physiotherapist were noted in 74%, 23%, and 14% of women, respectively. Fourteen percent (n = 12) of patients reported having undergone a surgical neovaginoplasty.

DISCUSSION

This is a large cohort descriptive report demonstrating the frequency of presenting symptoms and diagnostic/referral care in individuals with MRKH. The percentage of renal anomalies in our population, 19%, was lower than the prevalence of renal anomalies in other studies, 30%–50%, of women with MRKH (2, 4). The nonsurgical approach is the preferred approach based on the Dutch guidelines because there is no clinically relevant improvement in sexual function, there are limited complications of dilation therapy, and extensive dilation is also needed after surgical treatment (1). Regardless of the approach for neovaginoplasty, the use of multidisciplinary care is likely to benefit all women with MRKH syndrome. The investigators emphasize the importance of effective collaboration in a multidisciplinary team consisting of gynecologists, psychologists, sexologists, physiotherapists, and patient platform(s) in providing comprehensive care women with MRKH syndrome. Women with this rare and impactful condition may benefit from specific expertise because a significant proportion of women have a concomitant congenital condition, psychological distress, and/or dyspareunia (2, 4, 5). Moreover, the range of therapeutic options, including challenging surgical procedures, necessitates specific expertise.

As per guidelines, at Radboudumc, multidisciplinary care is provided to women with MRKH syndrome. Half to two thirds of the women who were provided such multidisciplinary consultations actually made use of this option. In addition to consultation with the gynecologist, this includes a standard offer of a consultation with a clinical psychologist and the opportunity to access peer-to-peer support (1). If deemed valuable, a referral to a sexologist or physiotherapist is provided. More than one third of women did not use sexology treatment provided. This is in spite of the fact that women with MRKH syndrome experience more pain during intercourse and lower sexual esteem and have a more negative genital self-image than those without the syndrome (5). It may still be embarrassing to bring up sexual issues, and low expectations of the effectiveness of treatments for sexual and pelvic floor problems may also play a role. The reported use of peer-to-peer support as described in the medical files (1 in 3 women, 35%) seems an underreport based on our clinical experience and information from the patient platform.

CONCLUSION

With the present study, we provide insight into the multidisciplinary care for women with MRKH syndrome at our center over the past 10 years. We found that the vast majority, but not all women, received such structured and comprehensive care at our center and only half of the women used multidisciplinary options that were provided. Conducting the first and, when needed, consecutive consultations together (gynecologist and psychologist) may help to make care more accessible and reduce barriers to seeking support. To present the sexologist as a regular member of the team may also improve the uptake of the options. We furthermore believe that correct timing and alignment with the women's needs are of paramount importance in delivering multidisciplinary care. Dissemination of results and experiences as described in this research letter, and feedback to referring colleagues on the basis of the existing guidelines, may improve the quality and continuity of care for this population.

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L.M. and S.J.L. should be considered similar in author order.

* In this paper we use the term "women with MRKH syndrome". Although we are aware that not all persons with MRKH syndrome identify as women, we use the term for reasons of brevity and consistency.

TABLE 1

Characteristics, diagnostics, and treatment of 85 women with Mayer-Rokitansky-Küster-Hauser syndrome treated at Radboud University Medical Center between 2012 and 2021.

Variable	No. of women with data available n (%)	Outcome in median (range) or number (percentage) as appropriate
Age at pubarche, y (range)	40 (47%)	11 (8–14)
Age at thelarche, y (range)	44 (52%)	12 (8–14)
Age at diagnosis, y (range)	85 (100%)	16 (12–29)
Vaginal depth at the first consultation, cm (range)	45 (53%)	3.0 (1.0–8.0)
Associated extragenital abnormality		
Renal malformation (%)	85 (100%)	16 (19%)
Skeletal malformation (%)	85 (100%)	10 (12%)
Hearing defect (%)	85 (100%)	9 (11%)
Limb abnormality (%)	85 (100%)	4 (5%)
Heart defect (%)	85 (100%)	2 (2%)
Sexual relationships		
Romantic relationship current or past (%)	83 (98%)	60 (72%)
Sexually active (%)	81 (95%)	55 (68%)
Penetrative sex (%)	79 (93%)	44 (56%)
Dyspareunia (%)	45 (53%)	21 (47%)
Sexarche, y (range)	23 (27%)	17 (13–26)
Age of the first penetrative sex, y (range)	21 (25%)	18 (13–26)
Main reasons for the first consultation		
Primary amenorrhea (%)	79 (93%)	46 (58%)
Cyclic pain (%)	79 (93%)	6 (7%)
Primary amenorrhea and cyclic pain (%)	79 (93%)	2 (3%)
Vaginal symptoms/dyspareunia (%)	79 (93%)	3 (4%)
Primary amenorrhea and vaginal symptoms (%)	79 (93%)	1 (1%)
Questions about treatment (%)	79 (93%)	11 (14%)
Request for follow-up/patient policy (%)	79 (93%)	2 (2%)
Questions concerning fertility (%)	79 (93%)	1 (1%)
Confirming diagnosis (%)	79 (93%)	2 (2%)
Other (%)	79 (93%)	5 (6%)
Diagnostics		
Diagnosis in primary care	82 (96%)	3 (37%)
Diagnosis in secondary care	82 (96%)	59 (72%)
Diagnosis in tertiary care	82 (96%)	20 (24%)
Vaginal examination	85 (100%)	56 (66%)
Hormonal tests	85 (100%)	57 (67.1%)
Genetic tests	85 (100%)	29 (34.1%)
MRI scan	77 (91%)	71 (92%)
Multidisciplinary care		
Psychologist present in the first consultation	85 (100%)	2 (2%)
Psychological questionnaire used	67 (79%)	8 (12%)
Consultation with a psychologist offered	85 (100%)	63 (74%)
Consultation with a psychologist	78 (92%)	40 (51%)
Consultation with a sexologist offered	85 (100%)	20 (23%)
Consultation with a sexologist	79 (93%)	12 (15%)
Consultation with a pelvic physiotherapist offered	85 (100%)	12 (14%)
Consultation with a pelvic physiotherapist	66 (78%)	7 (9%)
Peer-to-peer support offered	85 (100%)	72 (85%)
Peer-to-peer support used	55 (65%)	19 (35%)
Dilator treatment (N = 49)		
Age at start of dilator treatment, y (range)	43 (88%)	17.0 (14.0–39.0)

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TABLE 1

Continued.

Variable	No. of women with data available n (%)	Outcome in median (range) or number (percentage) as appropriate
Duration dilator treatment, mo (range)	36 (73%)	7.5 (1.0–84.0)
Vaginal depth before dilator treatment, cm (range)	31 (63%)	2.0 (1.0–6.5)
Vaginal depth after 6 mo, cm (range)	15 (31%)	6.0 (1.0–12.0)
Vaginal depth after 12 mo, cm (range)	8 (16%)	6.0 (3.0–10.0)
Surgery treatment (N = 12)		
Age at time of surgical procedure, y (range)	11 (92%)	19 (17–29)
Davydov procedure, N (%)	12 (100%)	9 (75)
Vecchietti procedure, N (%)	12 (100%)	3 (25)
Intraoperative complications (%)	12 (100%)	0 (0)
Postoperative complications (%)	12 (100%)	4 (33.3%) (Davydov, 2; Vecchietti, 2)[a]
Reoperation (%)	12 (100%)	3 (25%) (Davydov, 3)[b]
Vaginal depth before surgery, cm (range)	7 (58%)	2.0 (2.0–3.5)
Vaginal depth 6 months after surgery, cm (range)	5 (42%)	7.0 (5.0–12)
Vaginal depth 12 mo after surgery (cm)	6 (50%)	6.5 (4.0–10.0)

Note: MRI = magnetic resonance imaging.
[a] The types of postoperative complications were stenosis of the vagina (n = 2 both after Davydov), urinary tract infection (n = 1 after Vecchietti), and tingling sensation in 1 leg and severe headache without a neurological explanation in the same patient (n = 1 after Vecchietti).
[b] Reoperations were laparoscopic removal of a uterine rudiment, coagulation of neovaginal granulation tissue, and deepening of the vagina.

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CRediT Authorship Contribution Statement

Lisanne Martens: Writing – review & editing, Writing – original draft, Methodology, Formal analysis, Data curation, Conceptualization. **Susanna J. Lodewijk:** Writing – review & editing, Writing – original draft, Methodology, Formal analysis, Conceptualization. **Esther Leuning:** Writing – review & editing, Methodology, Data curation. **Anke J.M. Oerlemans:** Writing – review & editing, Supervision, Methodology, Conceptualization. **Chris M. Verhaak:** Writing – review & editing, Supervision, Methodology, Data curation, Conceptualization. **Kirsten B. Kluivers:** Writing – review & editing, Supervision, Methodology, Data curation, Conceptualization.

Declaration of Interests

L.M. has nothing to disclose. S.J.L. has nothing to disclose. E.L. has nothing to disclose. A.J.M.O. has nothing to disclose. C.M.V. has nothing to disclose. K.B.K. has nothing to disclose.

Lisanne Martens, M.D.[a]
Susanna J. Lodewijk[b]
Esther Leuning[c,d]
Anke J.M. Oerlemans, Ph.D.[b]
Chris M. Verhaak, Ph.D.[c]
Kirsten B. Kluivers, M.D., Ph.D.[a]

[a] Department of Obstetrics and Gynecology, Radboud University Medical Center, Nijmegen, the Netherlands; [b] IQ Health, Radboud University Medical Center, Nijmegen, the Netherlands; [c] Department of Medical Psychology, Radboud University Medical Center-Amalia Children's Hospital Nijmegen, Nijmegen, the Netherlands; [d] Karakter Child and Adolescent Psychiatry University Center, Nijmegen, the Netherlands

Correspondence: Lisanne Martens, M.D., Department of Obstetrics and Gynecology, Radboud University Medical Center, Geert Grooteplein Zuid 10, 6525 GA Nijmegen, the Netherlands.
E-mail address: L.Martens@Radboudumc.nl

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