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CASE REPORT

A TRAUMA PERSPECTIVE ON A CASE OF MAYER-ROKINTANSKY-KUSTER-HAUSER SYNDROME PRESENTING WITH MAJOR DEPRESSION WITH PSYCHOSIS

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ABSTRACT

The psychological impact of the diagnosis of congenital anomalies is an area with limited research; and more limited still when such congenital anomalies are not very obvious. Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome can be one of those less apparent congenital anomalies. Owing to their inability to bear children, women confronted with the diagnosis of MRKH can be left stressed, or worse, traumatized by the fact that they can no longer function in their traditional gender roles. This may leave any individual wanting to deny the diagnosis or even withhold such information. And the experience is a fertile ground for the development of any mental illness. MRKH occurs in every 1 out of 4000-5000 females. Although the psychological impact of stress is a well –established fact, there are fewer literature on the possible genetic link of MRKH and mental illness, the psychodynamic underpinnings and the similarities to trauma of the diagnosis of MRKH, in particular.

We present here a case of MRKH syndrome with an initial presentation of major depression and psychosis; with the diagnosis of MRKH initially withheld from the resident psychiatrist. We would also want to highlight a trauma perspective of the case presented as well as to emphasize the need for more research congenital anomalies are accompanied by co-morbid psychiatric illnesses.

KEYWORDS: *Uterine agenesis, Mullerian agenesis, trauma perspective, depression & psychosis, psychological distress*

CASE PRESENTATION

Psychiatric History

A 23-year-old single female, presented at the outpatient unit with depressed mood, insomnia, occupational impairment, hallucinatory gestures, paranoia, and suicidal ideations. The patient already had persistent amenorrhea and being from a low- socioeconomic strata, she had not prioritized consultation with an obstetrician. The social pressure of being in a relationship, getting married and having children also made her worry of her amenorrhea. When she was finally gainfully employed, she decided to consult an obstetrician about 3 ½ months ago.

At the time, she was found to have a congenital absence of the uterus and an ovary. The findings devastated her, enough to cause difficulty sleeping, depressed mood most of the day and impairment in occupational functioning. She hid the obstetric findings from anyone but she felt like people from work were talking about her and her obstetric findings.

Due to persistence of depressed mood, insomnia and suspiciousness of others, the patient was eventually convinced by her friends to consult a psychiatrist 2 ½ months ago, who initially gave her Escitalopram 10mg once at night, Quetiapine 300mg twice a day and additionally, Chlorpromazine 50mg at night due to persistent insomnia. However, she eventually discontinued the medications due to somnolence.

She consulted a different psychiatrist who gave her Fluoxetine 20mg once a day, Risperidone 1mg at night and Biperiden 2mg once a day – which she also discontinued due to slowing of her movements. The patient withheld the obstetric findings from the two psychiatrists she consulted. The patient originally rented an apartment near her place of work. But the nationwide lockdown due to the COVID-19 pandemic forced the patient to work remotely from her provincial home with her parents. She still felt depressed, with low energy and was socially withdrawn during this time. She became more and more suspicious of others talking about her and her obstetric findings.

Her symptoms worsened about a month ago, when she started neglecting her hygiene and her paranoia extended to people at church. . The patient's mother eventually found out about her initial obstetric finding and accompanied her to a different obstetrician, who had the same finding – of uterine agenesis or Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. This event worsened her depression and she became further withdrawn. The patient's mother restarted her last medications (Fluoxetine and Risperidone) but her depressive symptoms persisted, now with intermittent auditory hallucinations.

Despite this, the patient went back to her apartment with intentions of going back to work 1 week ago. However, she had an altercation with her landlady due to unsettled rental fees, resulting in incessant crying, hopelessness and worthlessness. To her friends, she verbalized her plan of overdosing with her medications. This eventually prompted consultation at the outpatient unit of our psychiatric hospital.

Past psychiatric history showed 2 episodes of major depression, both caused by academic stressors. Both also manifested similarly to the current episode: primarily with depressed mood, insomnia, low energy, suspiciousness and auditory hallucinations. Both episodes were also treated with the same medications: Escitalopram and Quetiapine, with the more current episode with an increased dose of Quetiapine. The patient, however, was poorly adherent to treatment and discontinued the medications when she started feeling better – around 3-4 months from medication initiation.
Family history showed mental illness from her paternal grandaunts and granduncle, manifesting mainly with psychotic symptoms.

Psychosocial history showed a complicated family dynamic fraught with financial problems, and strained relationship between the family and her relatives. The patient's paternal relatives were particularly enmeshed in familial concerns, financial and otherwise; so much so that academic pressure from them was enough to cause the patient's past 2 episodes of major depression. The paternal relatives provided the main financial support to the family. Her family is often labeled by her paternal relatives as a 'burden' to them – always reminding them of various expenses such as educational or medical expenses.

Objective Findings

The patient was seen at the outpatient clinic with a slim body and average height. She had secondary female sexual characteristics. She was behaved and had fair eye contact. She kept her arms crossed. During the interview she was noted with frequent glances towards her parents. She was responsive with hypo-productive speech, evasive and answered mostly with close-ended. She had depressed mood with congruent affect and expressed feelings of worthlessness and fear that this sadness will not go away. Noted with thoughts of suicide and a plan to overdose herself. She admitted having paranoid thoughts of other people talking about her. She mentioned feeling offended that she was brought to a psychiatric hospital upon the insistence of her mother. She had fair impulse control and intellectual insight with poor judgment. During the initial consultation, she withheld the information regarding her obstetric condition.

Physical examination was unremarkable with secondary sexual characteristics consistent with the female gender. No facial hair noted with developed mammary glands, wide hips and a small waist line.

Neurologic examination was also unremarkable. Diagnostic tests done such as complete blood count, urinalysis, blood chemistries, chest x-ray and ECG were all unremarkable. The patient withheld the information regarding her obstetric condition on the initial encounter but on subsequent follow-up, ultrasound findings showed "uterine agenesis with the presence of an ovary, right with left ovary not seen and fecal residue".

Beck's Depression Inventory (BDI) and Beck's Anxiety Inventory (BAI) showed the following results: 36 (Severe Depression) and 31 (Moderate Anxiety), respectively.

Diagnosis

Although the psychiatric diagnosis of the patient is rather direct and usual, the diagnostic challenge here is due in part to the withholding-information behavior of the patient. As the primary precipitant was withheld for a considerable amount of time, this posed an obstacle to achieving psychotherapeutic objectives.

A diagnosis of Major Depressive Disorder (MDD) with Psychotic Features (based on the DSM-5) was given; with an additional diagnosis of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, Type 1.

Therapeutic Intervention

The immediate goal of treatment for the patient was stabilization and safety because of her symptoms, mainly having suicidal ideations. The patient was advised admission but was rejected by the parents despite the risk.

Instead, pharmacologic intervention was employed together with psychoeducation and strict suicide precautions. Initially, the patient was started on Escitalopram 10mg at night, Clozapine 50mg at night and Biperiden 2mg as needed for extrapyramidal symptoms. Close follow-up was advised, but the patient returned for consultation after a month.

On her first follow-up, the patient eventually disclosed her obstetric condition, citing this as a the main stressor precipitating her depression. Pharmacologic intervention with psychological support and extensive psychoeducation was done.

The succeeding 6 months were fraught by difficulties due to the pandemic. Follow up consultations were shifted to virtual consultation due to rising cases of COVID-19. The patient was initially poorly adherent to the medications, often missing doses or not returning for scheduled follow-up. Depressive symptoms persisted and the patient eventually quit her work. Anxious distress became more apparent, with suicidal thoughts and depression with mixed features.

Medications were eventually adjusted to Escitalopram 20mg/day, Lithium Carbonate 450mg/day and Diphenhydramine 50mg for insomnia. Succeeding follow-ups showed fluctuations in mood. For a time, the patient became temporarily employed again.
Psychological support and counseling were also given during follow-up visits, especially because of her MRKH. On her most recent follow-up, she was on Escitalopram 20mg/day, Lithium Carbonate 450mg/day and Quetiapine 12.5mg as needed for insomnia.

CASE DISCUSSION: The Trauma of MRKH

Relation between stress and mental illness is a well- established fact. Of course, mental health issues are common given a congenital, chronic or untreatable illness – especially one that is uncommon and not very apparent. Case reports (which most papers on MRKH are) of mental disorders precipitated by the diagnosis of MRKH were found reporting depression (1), acute psychosis (2), and even dissociative disorders (3). The diagnosis of MRKH is viewed here as a traumatic event – one that threatened deeply held expectations of life. Therefore, keeping in mind this unconventional view of trauma, the case above is being presented.

Uterovaginal/uterine agenesis, Mullerian agenesis or Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital malformation characterized by the absence of uterus and the upper two-thirds of the vagina, with an incidence of 1 in 4000-5000 females (4).

In typical cases (MRKH type 1), ovaries are unaffected but rarer atypical cases are also possible where there is unilateral or bilateral ovarian agenesis (5). Primary amenorrhea seems to be the most common presenting symptom and MRKH is an incidental finding. There are few studies on MRKH and fewer still explore its psychological impact.

But definitely, adjusting to the diagnosis of MRKH is a difficult and traumatic process for all women, leading them to question their identity as women and to experience a sense of confusion regarding their gender, their bodies, as well as their social and sexual roles.

It is clear in this case that the precipitating stressor for the development of depression with psychotic features was the diagnosis of MRKH. Withholding such information from her family, while not clearly understanding the diagnosis or maybe forgetting details about it, is reminiscent to how most patients react when confronted with a new diagnosis of cancer.

The initial diagnosis of MRKH was indeed a traumatic experience. Oftentimes, memory for "bad news consultations" is fragmented and not organized in a coherent narrative, with misconceptions about what was said and what was wrong (6)

This is on a continuum with what happens in the development of PTSD, where trauma memory is poorly elaborated and not incorporated fully into the autobiographical memory base (7).

Being given a diagnosis of the "absence of a vagina and uterus" is difficult to comprehend. The uterus – thus, the ability to bear children is typically a fundamental assumption of not only a function, but also one's identity.

From a Kohutian perspective, a continuous, stable sense of self or identity – one crucial aspect of narcissistic organization (8), is often threatened upon diagnosis (9). Thus, furthering the development of depression.

A cognitive behavioral model can also be used to try and understand the patient's reactions to different situations. For many young women with MRKH, their body schema, i.e. a woman's internal mental representation of her body on which – according to Piaget and Cook (10) – all other schemata are built, is shattered. Precipitating her desire to get checked because of amenorrhea was the social pressure of her friends getting married and having children. After the diagnosis of MRKH, the patient might have negatively appraised herself as being left out, inferior, or defective as a woman (11). Seeing how the patient's paternal relatives are enmeshed with personal matters and negatively seeing herself as a burden or even an embarrassment to the family might have caused her to withhold the information from her family (and everyone else) or even disregard further diagnostic tests as a maladaptive attempt to control (or even deny) the sense of threat to her body and her womanhood. However, this can result to feelings of isolation, further helplessness and even delay possible management strategies.
Hence, these psychodynamic underpinnings not only makes the diagnosis of MRKH a threat to her self -esteem and a traumatic blow to her sense of self but also makes her reactions reminiscent of those with PTSD. However, there is a key difference between the psychological reactions to MRKH and PTSD. In PTSD, the sense of current severe threat is, above all, maintained psychologically through the nature of the

trauma memory, the resulting negative appraisals, and the strategies intended to control threat or symptoms (12). In contrast, in MRKH, in addition to these processes it is the unalterable presence of the actual physical impairment that underpins and feeds into the sense of current threat (7).

Genetic Link between MRKH and Mental Illness
With the emotional burden accompanying the diagnosis of MRKH alone, it is expected that many individuals would encounter some form of psychological stress.

In 2020, Chen, et al. did an analysis of 141 cases of MRKH and found that a total of 75.2% of MRKH patients suffered from depressive symptoms, and 34.0% reached a moderate to severe level (1). There are less studies on the genetic link of MRKH with mental illness in general but one study showed that microdeletion of 17q12 can be a cause for MRKH. This microdeletion also carries a significant risk for cognitive impairment, autism and even schizophrenia (13).

Although this is not present for certain in this case presented, this warrants further research in the genetic link of MRKH or other congenital mental illnesses.

The patient initially withheld information from her family and also from the psychiatrist evaluating her. Withholding information behavior (WIB) is fairly common when stigma and embarrassment is associated with an illness.

Medical literature recognizes that quality communicative interactions between patients and their doctors are important to the certainty of diagnosis and success of treatment. Limited disclosure of critical health information serves as a barrier to this. Despite this intuitive knowledge, there is evidence that suggests patients purposefully withhold relevant information from clinicians (14).

Psychiatrists should stay flexible in communicative interaction with patients, from asking close- to open-ended questions, keeping the interview conversational or staying objective when need be. However, despite this, withholding information behavior (WIB) can still happen. Studies would show that more depressive symptoms are associated with increased WIB and of course the stigma and the perceived embarrassment associated with MRKH affects this as well (15).

Fortunately, studies also show that status of patient-physician relationship (PPR) is significantly associated whether patients withhold information from their doctors. This reinforces the support for interventions bolstering the PPR (15).

Psychotherapy

Employing self-psychology can be an approach to psychotherapy. Empathy can be utilized as a therapeutic end in itself. A synthesis & articulation of the patient's inner world, thoughts and feelings, will enable the patient to be deeply understood and accepted. Cumulative trauma destabilizing prior narcissistic balance may be alleviated through validating and affirming mirroring of the patient's experience. Integrating self-psychology with attachment theory may also be utilized. Empathic understanding promotes a secure base, allowing self-regulation, more robust self-esteem and more secure attachments (9).

Through a cognitive behavioral model, the following steps can be utilized for psychotherapy: revisit the experience of being diagnosed and help them to fully process what has happened and integrate it; examine self-appraisals and beliefs and help them to challenge and modify these; develop a more accepting view of their MRKH as something that is no longer a shameful secret; and, address any maladaptive coping strategies that may serve to maintain unhelpful self-beliefs (7).

CONCLUSION
The above case is a stark representation of how a rare, non-apparent congenital anomaly can affect one's sense of being. The diagnosis of MRKH was a severe traumatic blow to the patient's sense of womanhood, effectively causing depression. Common symptoms of depression, manifesting in this patient, may result in utilizing the usual therapeutic approach to depression - effectively putting this patient in a box. Let us not forget that much advancement

is occurring in the realm of psychotherapy and psychiatry as a specialty; and we should not be limited to what we already know. The case presented in this paper is viewed through a different lens - that of a trauma perspective.

Much research is needed in terms of MRKH and its psychological impacts. Screening for medical illnesses during the psychiatric interview would minimize the possibility of missing out on such information; and thus allow the psychiatrist to manage such patients more holistically.

REFERENCES

1.Chen N, Song S, Duan Y, Kang J, Deng s, Pan H, Zhu L. Study on depressive symptoms in patients with Mayer-Rokitansky-Küster-Hauser syndrome: an analysis of 141 cases. Orphanet J Rare Dis. 2020 May 24; 15:121. Available from: https://doi.org/10.1186/s13023-020-01405-9

2.Nath K, Boro B, Naskar S. A Rare Case of Mayer-Rokitansky-Kuster-Hauser Syndrome Presenting with Acute Psychosis. Journal of clinical and diagnostic research. 2016; 10(4): VD03–VD04.

3.Joshi S, Bhandari AR, Shrestha P, Shakya R. Dissociative disorder in Mayer Rokitansky Küster Hauser syndrome with pulmonary agenesis: a case report. Egypt J Neurol Psychiatry Neurosurg. 2022; volume 58: number 3. Available from: https://doi.org/10.1186/s41983-021-00440-y

4.Herlin M, Bjørn A-MB, Rasmussen M, Trolle B, Petersen MB. Prevalence and patient characteristics of Mayer–Rokitansky–Küster–Hauser syndrome: a nationwide registry-based study. Hum Reprod. 2016 Oct;31(10):2384–90.

5.Oppelt P, Renner SP, Kellermann A, Brucker S, Hauser GA, Ludwig KS, et al. Clinical aspects of Mayer–Rokitansky–Kuester–Hauser syndrome: recommendations for clinical diagnosis and staging. Human Reproduction. 2006 March;21(3): 792–797.

6. Fallowfield L, Jenkins V. Communicating sad, bad, and difficult news in medicine. Lancet. 2004 Jan 24;363(9405):312-9.

7. Heller-Boersma JG; Schmidt UH, Edmonds DK. A Cognitive Behavioural Model and Therapy for Utero-Vaginal Agenesis (Mayer-Rokitansky-Kuster-Hauser ¨ Syndrome: MRKH) Behavioural and Cognitive Psychotherapy. 2009 Jul;37 (4): 449–467. Available from: doi:10.1017/S1352465809990051

8. Stolorow RD, Lachmann FM. Psychoanalysis of developmental arrests: Theory and treatment. International Universities Press Inc; 1980.

9. Leon IG. Understanding and Treating Infertility: Psychoanalytic Considerations. Journal of The American Academy of Psychoanalysis and Dynamic Psychiatry. 2010; 38(1): 47–75.

10. Piaget J & Cook MT. The origins of intelligence in children. New York, NY: International University Press; 1952.

11. Heller-Boersma JG, Schmidt UH, Edmonds DK. Psychological Distress in Women With Uterovaginal Agenesis (Mayer-Rokitansky-Küster-Hauser Syndrome: MRKH). 2009; 50(3): 0–281.

12. Ehlers A, Clark DM. A cognitive model of posttraumatic stress disorder. Behav Res Ther. 2000 Apr;38(4):319-45

13. Hinkes B, Hilgers KF, Bolz HJ, Goppelt-Struebe M, Amann K, Nagl S, et al. A complex microdelaetion 17q12 phenotype in a patient with recurrent de novo membranous nephropathy. BMC Nephrol. May 2012; volume13: Number 27. Availble from: https://doi.org/10.1186/1471-2369-13-27

14.Sjöstrand M, Sandman L, Karlsson P, Helgesson G, Eriksson S, Juth N. Ethical deliberations about involuntary treatment: interviews with Swedish psychiatrists. BMC medical ethics. 2015 May 28;16: 37.

15. Yang X, Parton J, Lewis D, Yang N, Hudnall M. Effect of Patient-Physician Relationship on Withholding Information Behavior: Analysis of Health Information National Trends Survey (2011-2018) Data. J Med Internet Res. 2020 Jan 29;22(1):e16713

BROKEN VESSEL: EXPLORING THE LOSS OF WOMANHOOD AND GRIEVING THE ABSENT UTERUS

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For the final interview round of Miss Universe 1994, then Miss India Sushmita Sen was asked: What for you is the essence of being a woman?, to which she replied: “Just being a woman is a gift of God that all of us must appreciate. The origin of a child is a mother, who is a woman. She shows a man what caring, sharing and loving is all about. That’s the essence of being a woman.” She went on to win the Miss Universe title that night along with the hearts of millions of viewers.

Most of us have heard this question: “What is the essence of being a woman?”; and I’m sure most of us would, automatically or perhaps inadvertently, relate it to motherhood. For centuries, millennia even, girls across different cultures were brought up and primed to eventually embrace and even yearn for motherhood, which remains true until now.

This society that we live in, enumerates what makes a person valuable. For a woman in this society, what makes her worthy are not enumerated but numbered and limited. Female limitations have been ingrained since one’s childhood that as adults are internalized. And just like half of the artwork here – the woman is boxed in by certain societal expectations as indicated by the long flowing hair, ample breasts and most importantly – her fertile womb.

But what if, a woman wanted to shave her head, just like the other half of this artwork, because she believes it suits her. Or had one of her breasts removed because this could be life saving, or she was born without a uterus, without a womb.; without the very organ for which she is valued. This is the struggle that many women face in their daily lives as they interact with various people in their lives, but the most difficult battle actually stems from within.

Imagine then what it would be like if you were deemed a woman all your life then one day you no longer fulfill the criteria for being a woman. How traumatic it could be when your own expectations in life are forever shattered. This then leads you to question your own identity.

The artwork is not just a mere contrast of 2 different perspectives of womanhood but more so an abrupt transition from one to the other, accompanied by a sudden and different kind of trauma. This artwork is a staunch representation of our case.