Staging and clinical diagnosis in Mayer-Rokitansky-Küster-Hauser syndrome

Peter Oppelt

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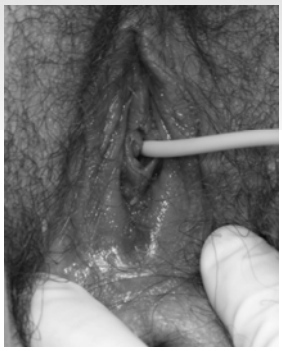
The First Impression



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Introduction

- Clinically, this malformation of the female genital organs presents:
  - a rudimentary solid bipartite uterus with
  - solid vagina
  - Sometimes associated malformations
    (uterus bipartitus solidus rudimentarius cum vagina solida)

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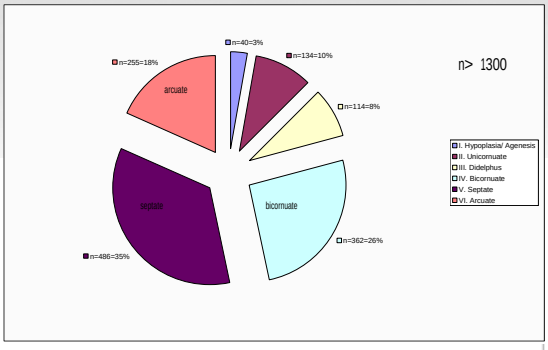
Distribution of Uterine Malformations



Woelfer and Rimbach (2005) Gyn Endokrin 3: 18 – 24

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Causes of uterine malformations

- Failure of the development or incomplete fusion of Mullerian ducts - the facilities of the uterus, cervix and upper vagina
- Period between the 10th and 17th embryonic weeks
- Exact causes for this malformation are still not known
- Familial clusters often observed

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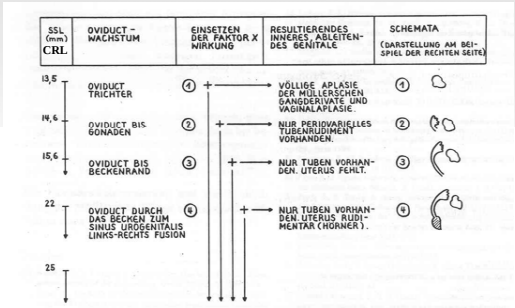
Manifestation of malformations according to time of pregnancy



Schmid-Tannwald I, Hasser GA Gebfra 37: 386, 1977

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First descriptions of “MRKH”

- 1829 Mayer - Bonn anatomist and physiologist Mayer
- 1938 Rokitansky – reported one case
- 1910 Kuester - first time summarized and collected individual cases from the literature in a review paper
- 1961 Hauser - 21 Case Descriptions on live patients (MRKH-Syndrome)

Classification of

AFS

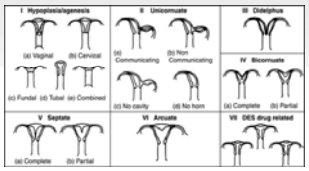

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Table I. Clinical and embryological classification of the malformations of the female genital tract (modified from Acién, 1992).

1. Agenesis or hypoplasia of a whole urogenital ridge: Unicornuate uterus with uterine, tubal, ovarian and renal agenesis on the contralateral side.
2. **Mesonephric anomalies** with absence of the Wolffian duct opening to the urogenital sinus and of the ureteral bud sprouting (and therefore, renal agenesis). The 'inductor' function of the Wolffian duct on the Müllerian duct is also failing and there is usually:
Utero-vaginal duplicity plus blind hemivagina ipsilateral with the renal agenesis, clinically presented as:
a) Large unilateral hematocolpos*
b) Gartner's pseudocyst on the anterolateral wall of the vagina*
c) Partial reabsorption of intervaginal septum, seen as a 'buttonhole' on the anterolateral wall of the normal vagina which allows access to the genital organs on the renal agenesis side.
d) Vaginal or complete cervico-vaginal unilateral agenesis, ipsilateral with the renal agenesis, and (1) with no communication, or (2) with communication between both hemiuteri (communicating uteri).
3. Isolated **Müllerian anomalies** affecting:
a) Müllerian ducts: they are the common uterine malformations as unicornuate (generally, with uterine rudimentary horn), bicornuate, septate and didelphys uterus.
b) Müllerian tubercle: cervico-vaginal atresia and segmentary anomalies such as transverse vaginal septum.
c) Both, Müllerian tubercle and ducts: (uni- or bilateral)
Mayer-Rokitansky-Kuster-Hauser syndrome.
4. Anomalies of the urogenital sinus: cloacal anomalies and others.
5. Malformative combinations: Wolfian, Müllerian and cloacal anomalies.

*These types can associate a vaginal ectopic ureter and interseptal or interuterine communication.

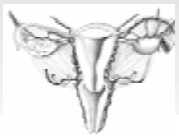
Classification of MRKH

MRKH syndrome	Associated malformations
Typical	Tubes, ovaries, and renal system generated and developed
Atypical	Malformations in the ovary or renal system
MURCS	Malformations in the skeleton and/or heart; muscular weakness, renal malformations

MURCS = Müllerian aplasia, renal aplasia, and cervicothoracic somite dysplasia (association).

Schmid-Tannwald/ Hauser 1977 & Duncan 1979

VCUAM-Classification



Vagina

Cervix

Uterus

Adnex

Malformations associated

"analogous to the TNM classification of oncological tumors "

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VCUAM-Classification

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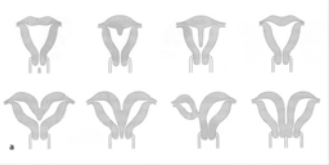
VCUAM-Classification

Uterus (U)	0	Normal
	1a	Arcuate
	1b	Septate <50% of the uterine cavity
	1c	Septate >50% of the uterine cavity
	2	Bicornate
	3	Hypoplastic uterus
	4a	Unilaterally rudimentary or aplastic
	4b	Bilaterally rudimentary or aplastic
	+	Other
	#	Unknown



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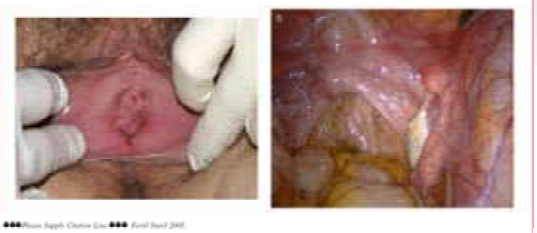
Classification of the Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome

FIGURE 3

VCUAM classification of atypical Mayer-Rokitansky-Küster-Hauser syndrome: V5b, C2b, U4b, A0, M0. (A) Vagina. (B) Intraoperative view.



Higher incidence of linked malformations in siblings of Mayer–Rokitansky–Küster–Hauser-syndrome patients

M. Wottgen[1], S. Brucker[2], S.P. Renner[1], P.L. Strissel[1], R. Strick[1], A. Kellermann[1], D. Wallwiener[2], M.W. Beckmann[1] and P. Oppelt[1,3]

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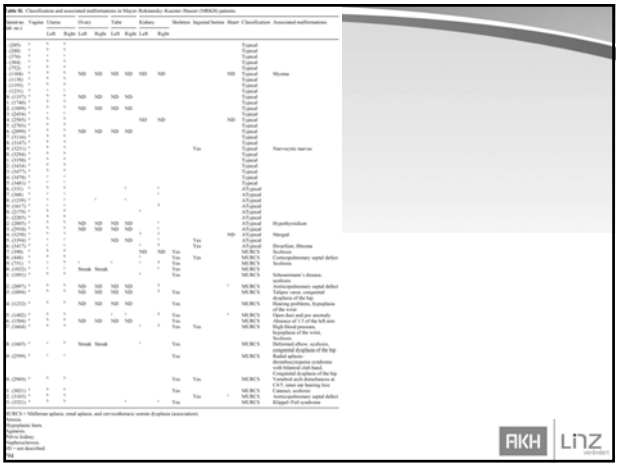
Table III. Comparison of the frequency of malformations in the present selected Mayer–Rokitansky–Küster–Hauser Group and in the normal population (Mainz population based birth Registry).

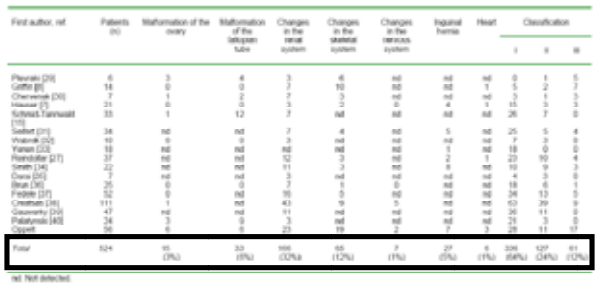
Organ system	Siblings of MRKH patients (n = 103, Erlangen + Tübingen)		Mainz Registry (n = 40 063) per 10 000		Pearson's chi-squared test
	n	%	n	%	
Musculoskeletal system	10[a]	7.76	237	2.37	$P < 0.005$
Cardiovascular system	3[b]	2.91	147	1.47	$P = 0.165$
Internal and external urogenital system	2[c]	1.94	154	1.54	$P = 1.0$

[a]Patient No.: 1252, 2179, 3047, 3048, 3258, 3294, 3304, 4230.
[b]Patient No.: 2097, 3116, 3258.
[c]Patient No.: 1617, 3881.



Assoziierte Fehlbildungen - MRKH

Table 3 Distribution of the associated malformations in the MRKH groups reported in the published literature (multiple descriptors per organ group were possible)



Oppelt et al. Clinical aspects of MRKH-Syndrome. Hum Reprod 03-2006

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Essential examinations

- Chromosome analysis
- [Diagnostic laparoscopy]
- Hormone status (LH, FSH, E2)
- Renal ultrasonography

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Recommended additional examinations

- Ultrasound of the vaginal vestibule, rectum
- MRI of the kidneys, small pelvis (or alternatively intravenous pyelography)
- Ovarian biopsy

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Principal symptoms for supplementary examinations to clarify associated malformations

Symptoms	Diagnostic clarification
Urinary incontinence	Urodynamics
Quick exhaustion	Myography, echocardiography
Skeletal malformations	Radiography, computed tomography if appropriate
Hearing loss	Audiography