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Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC): A Mental and Physical Agony in a Young: A Case Report

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Abstract: ***Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) is a rare autosomal dominant disorder characterised by multiple cutaneous and uterine leiomyomas, with an increased risk of developing aggressive renal cell carcinoma. This case study presents a 28yr old woman from a low socioeconomic background with a history of heavy menstrual bleeding, multiple fibroids, and infertility, leading to a total abdominal hysterectomy after unsuccessful myomectomy. A strong family history of fibroids was noted, with her mother and sisters experiencing similar conditions. Postoperatively, dermatological examination confirmed piloleiomyoma, and nephrological evaluation revealed early - stage renal cell carcinoma. This case highlights the importance of early diagnosis through clinical evaluation, pedigree analysis, and serial imaging. Increased awareness, genetic counselling, and further research are essential to improve patient outcomes and early detection of HLRCC.***

Keywords: HLRCC, uterine leiomyoma, renal cell carcinoma, piloleiomyoma, FH, genetic counselling, early diagnosis

1. Introduction

HLRCC is a rare autosomal dominant condition characterized by multiple cutaneous and uterine leiomyoma. These individuals are at an increased risk of developing the aggressive form of Renal Cell Carcinoma (1). It is also called as Reed's Syndrome named after Reed's and colleagues noticed an accumulation of the condition in 2 families with an autosomal dominant inheritance pattern. (4) It is caused by changes in the fumarate hydratase (FH) gene located on chromosome 1q43 that regulate an enzyme in the Kreb's cycle (2).

2. Case History

A 28yr old woman from low socioeconomic class came to the casualty with complaint of heavy menstrual bleeding since 45 days with mass per abdomen, weakness and multiple skin lesions over thighs, legs and trunk (Fig.2). The physical and mental agony started 4 years back with menorrhagia for one year after which, she was diagnosed with fibroid uterus and had undergone open myomectomy in February 2020, in which 35 myomas were removed. After one year, she got married and again after one year of marriage, she came with complaints of menorrhagia and infertility. She was diagnosed with bulky uterus with multiple fibroids in USG and HSG showing normal uterine cavity with bilateral cornual block. She was disappointed as her husband left her knowing this condition and took treatment only for menorrhagia with OCPs. One year later, she appeared in casualty with the above said condition.

She had a family history of her mother having multiple fibroid for which TAH (total abdominal hysterectomy) was done at the age of 35years.

All her three sisters had similar history of multiple fibroids and the elder sister underwent TAH at the age of 32years (Fig.1).

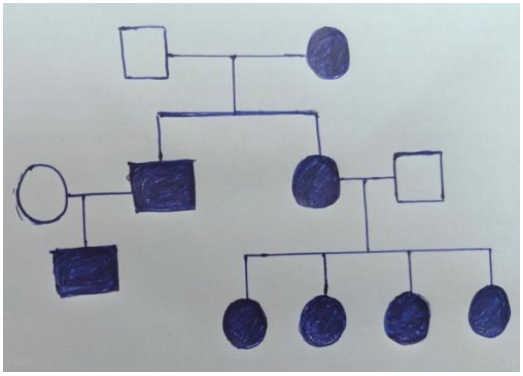

Figure 1

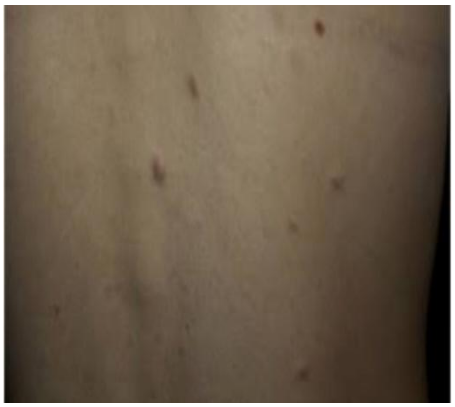

Figure 2

Investigations:
Hb - 3.9gm%, TPC - 2.78Lakh/microlitre
Blood Group - B positive
TLC - 4800/microlitre
HIV/HBsAg/HCV - Non reactive

Thyroid profile - WNL
RFT - Urea: 9mg/dl, Creatinine: 0.43mg/dl.
LFT - WNL
TUMOR MARKERS - CA 19 - 9: 7.6U/ml
LDH - 161.4IU/ml CA - 125 - 12.2U/ml
MRI (Abdomen and Pelvis) - Enlarged uterus (190mm*164mm*82 mm) with multiple SOLs (myoma) and largest measuring (83mm*53mm) and
Bilateral Hydroureteronephrosis
Peripheral smear –Microcytic hypochromic anaemia

Examination:

- **General Examination** - Patient conscious, oriented. BP - 100/70mmHg, PR - 90/min, pallor++
- Multiple nodular lesions over thigh, legs and trunk.
- **Systemic Examination** - CVS - S1S2 sounds heard, Murmur (-), RESP. SYS - Chest - clear, B/L VBS,
- GI: Liver, Spleen not palpable
- **P/A - Mass corresponding to 24week size uterus, firm, irregular, restricted mobility, lower pole could not be reached**
- **P/S** - Cervix taken up, no discharge, no mass
- P/V - Cervix taken up, uterus 24wk size, restricted mobility, B/L fornices full and nontender

Management:

First anaemia correction was done with 4 units of PRBC and calcium gluconate. Symptomatic treatment of menorrhagia was done with Norethisterone 15mg TDS. Open Myomectomy was planned under general anaesthesia in view of her fertility with consent for Hysterectomy if required. Intra - operative findings - The whole uterus and the uterine cavity was so distorted with almost more than 40 myomas. Hence, with the consent of patients' relative, Total Abdominal Hysterectomy with Bilateral Salpingectomy was done sparing the ovaries (Fig.3, 4). The patient was referred to a Dermatologist, where it was confirmed as Piloleiomyoma after biopsy. Due to early changes in kidney, i. e., Bilateral Hydroureteronephrosis, further the case was referred to a Nephrologist. After serial imaging and biopsy after 6 months, she was diagnosed as early stage of Renal Cell Carcinoma.

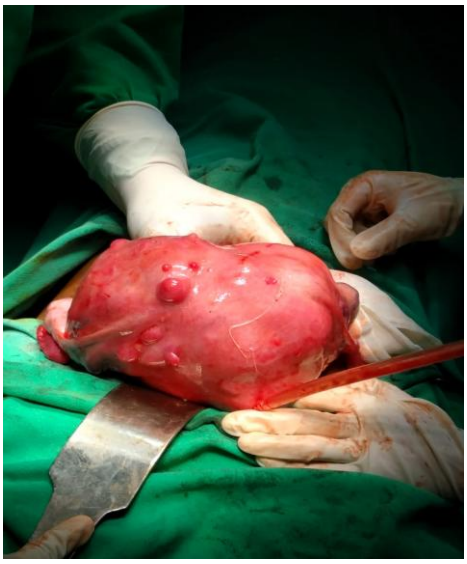

Figure 3

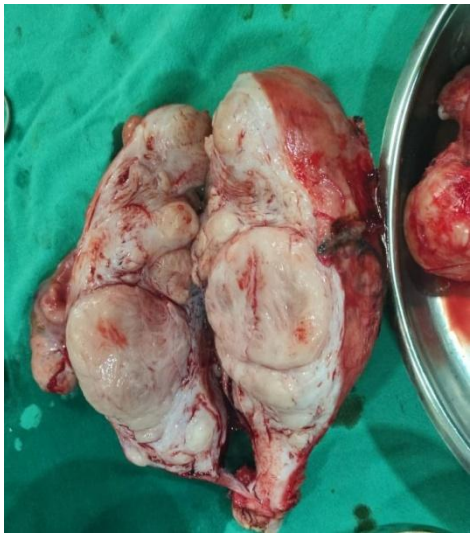

Figure 4

3. Conclusion

HLRCC is a rare autosomal dominant disorder with approximately 10 - 16% of them develop life threatening Renal Cell Carcinoma. i. e. Papillary RCC Though genetic typing is confirmatory tool but clinical diagnosis along with pedigree analysis and serial imaging can easily lead to the diagnosis of Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC)

Clinical criteria proposed for diagnosis of HLRCC:

1) Histologically confirmed multiple cutaneous piloleiomyomas.
2) At least 2 of the following manifestations: surgical treatment of uterine leiomyoma before age 40, type 2 papillary RCC before age 40 or a first degree family member who meets one of these criterias.

More researches, awareness and proper counselling are required for HLRCC.

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