

Herlyn-Werner-Wunderlich Syndrome의 새로운 진단기준 제안

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Herlyn-Werner-Wunderlich Syndrome: Extended Diagnostic Insights

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Backgrounds: Herlyn-Werner-Wunderlich syndrome (HWW) is rare congenital malformation syndrome characterized by a triad of uterus didelphys, blind hemivagina, and ipsilateral renal agenesis. This study reviews the imaging findings and suggests a scoring system for the diagnosis.

Methods: We collected 68 female patients with a clinical suspicion of HWW in our hospital. The ultrasonography and MRI findings were reviewed. We made a scoring system with regard to each one of the HWW triad as follows (Table 1); vaginal score (V1, vaginal septum with obstruction; V2, vaginal septum without obstruction; V3, no visible vaginal septum or vaginal atresia/agenesis), uterine score (U1, uterine didelphys; U2, bicornuate uterus; U3, septated uterus; U4, other uterine anomaly), and kidney score (K1, ipsilateral renal agenesis; K2, ipsilateral ectopic dysplastic kidney). Our extended diagnostic system consisted of three groups as follows; Group A: complete; Group B: incomplete; and Group C: probable HWW.

Results: The mean age at the diagnosis of HWW syndrome and current age were 16.1 and 21.8 years, respectively. The age at diagnosis were youngest in Group A compared to group B or C ($p=0.041$). Common symptoms related to the disease were mainly abdominal pain, dysmenorrhea and vaginal bleeding or frequent discharge. Of the 10 patients who have attempted a conception, 4 were infertile and 6 were reproductively normal. At median follow-up period of 32 months, renal function was evaluated in 61 patients (Table 5). While most (95.1%) maintained normal renal function, except for 2 patients with end-stage renal disease (ESRD) and one patient with chronic renal insufficiency (CRI). Urinary anomalies were frequently observed and ectopic ureter with insertion to the vagina or the bladder was the most common finding. Of significant note, ovarian tumor developed in five patients: 2 endodermal sinus tumor, 1 teratoma, and 2 serous cystadenoma.

Conclusions: Considering the phenotypic heterogeneity of HWW, early diagnosis in extended spectrum may screen out the over-looked population. Understanding the high incidence of comorbid conditions including malignancy potential and monitoring on the renal and fertility outcome are also of importance. Early referral to a nephrologist and multi-disciplinary care in longer term would improve the prognosis. Later identification of the underlying genetic defect may provide a better understanding of the genotype-phenotype correlation.

Key Words: 쌍각자궁, 단일신, 자궁기형

Herlyn-Werner-Wunderlich, Uterus didelphys, Renal agenesis