

## The effectiveness of Wilms tumor screening in Beckwith-Wiedemann spectrum (2019)

Alessandro Mussa, Kelly A. Duffy, Diana Carli, Jessica R. Griff, Riccardo Fagiano, Jonida Kupa, Garrett M. Brodeur, Giovanni Battista Ferrero, Jennifer M. Kalish

### Background

Patients with Beckwith-Wiedemann spectrum (BWS) have an increased risk for developing embryonal tumors. Wilms tumor (WT), a cancer of the kidney, is the most common tumor to develop. The risk for WT varies for patients with BWS dependent on the molecular cause. Patients with BWS due to gain of methylation at imprinting control region 1 (IC1 GOM) and patients with paternal uniparental disomy of chromosome 11p15 (pUPD) have the highest risk for WT and patients with BWS due to loss of methylation at imprinting control region 2 (IC2 LOM) have a low risk for WT. It is recommended that patients with BWS receive routine tumor screening by ultrasonography every 3 months until the age of 7 years to help identify WT early.

### Purpose

This study evaluated the ages at WT development in patients with BWS compared to the ages at WT development in patients without BWS (SEER registry patients) to determine whether screening until age 7 years is sufficient. The stage of WT at time of diagnosis was compared between patients with BWS diagnosed through screening and those who were not screened.

### Findings

Patients with BWS tend to be diagnosed with WT earlier than patients without BWS. More than one third (35.5%) of patients with BWS are diagnosed with WT by age 1 year (12 months). Almost 95% of patients with BWS are diagnosed by age 7 years (84 months), the age at which WT screening is discontinued. Although rare, patients with BWS have been diagnosed with WT older than the age of 7 years. The most common genetic types to develop tumors were patients with IC1 GOM and pUPD11, however some patients with IC2 LOM and 11p15 chromosome rearrangements were also diagnosed with WT.

Patients with BWS who were diagnosed through ultrasound screening were diagnosed at an earlier age and experienced a lower tumor stage at diagnosis compared to patients with BWS who did not receive screening. Patients with BWS who did not receive screening were diagnosed at similar ages to patients without BWS.

### Conclusion

Wilms tumor screening in patients with BWS is effective in reducing the stage at time of diagnosis. The current screening recommendations of ultrasound screening until age 7 years will identify approximately 95% of patients with BWS and WT.

### Key Points

- Patients with BWS are diagnosed with WT younger than patients without BWS.
- Screening is effective in identifying tumors early.

### Reference

Mussa A et al. The effectiveness of Wilms tumor screening in Beckwith-Wiedemann spectrum. *J Cancer Res Clin Oncol*. 2019;145(12): 3115-3123. PubMed PMID: 31583434