# Tumor screening in Beckwith-Wiedemann syndrome - to screen or not to screen? (2016)

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## Background

Patients with Beckwith-Wiedemann syndrome (BWS) are at an increased risk to develop tumors and receive tumor screening with the goal of early detection of tumors. Current recommendations suggest that all patients with BWS receive tumor screening. Tumor risk has been shown to differ by the molecular subtype of BWS. As a result, some groups in Europe proposed to change tumor screening guidelines according to BWS molecular subtype.

## Purpose

There are many challenges to changing tumor screening protocols. This invited comment discussed some of the related challenges and proposed to continue uniform screening for all patients with BWS.

## Findings

***Proposed Recommendation Changes***

It has been recommended that only patients with higher risks for tumor development should be screened. Patients with BWS due to loss of methylation at imprinting control region 2 (IC2 LOM) have a lower tumor risk (2.6%) and it was suggested that these patients should not receive tumor screening. It was also suggested that using alpha-fetoprotein (AFP) screening for hepatoblastoma is not necessary due to challenges with its interpretation and the invasiveness and anxiety that repeated blood draws can cause. Wilms tumor screening was suggested to discontinue after age 5 years.

***Implementation of Tumor Screening into Medical Practice***

Multiple factors influence how tumor screening protocols are implemented:

- Patient and societal financial considerations: Tumor screening is more cost effective than treatment of an undetected tumor.
- Parental and patient anxiety around screening: Screening can improve the emotional wellbeing of families by offering reassurance and a sense of control.
- Legal ramifications of missed diagnosis: Accessibility to care and legal liability varies from country to country. Europe typically uses a 5% risk threshold for determining which BWS subtypes should be screened. The United States uses a 1% risk threshold for tumor screening.

***Limitations of Proposed Changes***

- Access to genetic testing: Patients need to have access to genetic testing to know their genetic subtype and its associated tumor risk.
- Effectiveness of AFP screening: Regular AFP screening often detects hepatoblastoma development before an ultrasound can, leading to earlier diagnosis and better outcomes.
- Length of Wilms tumor screening: Further data on age of Wilms tumor (WT) development is needed before discontinuing WT screening after age 5 years.
- Implementation challenges: No consensus exists for clinicians to determine the best tumor screening recommendations and practices.
- Screening recommendations should consider that when there is no agreed upon consensus for appropriate tumor screening guidelines, it can be difficult for clinicians to implement screening.

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## Conclusion

Screening guidelines require consideration of cost-effectiveness of screening, patient and family anxiety, and clinical implementation techniques. All of these factors should be evaluated in order to determine the best approach to altering tumor screening in patients with BWS.

## Key Points

- Each genetic subtype of BWS carries a different risk for tumor development.
- Multiple factors should be considered to effectively alter tumor screening recommendations.
- Due to differing medical systems, screening recommendations may differ between countries.

## Reference

Kalish JM, Deardorff MA. Tumor screening in Beckwith-Wiedemann syndrome - To screen or not to screen? *Am J Med Genet A*. 2016;170(9), 2261-2264. PubMed PMID: 27518916