Children's Hospital of Philadelphia
RESEARCH INSTITUTE

Tumor screening in Beckwith-Wiedemann syndrome: parental perspectives (2017)

Kelly A. Duffy, Katheryn L. Grand, Kristin Zelley, Jennifer M. Kalish

Background

Children with Beckwith-Wiedemann syndrome (BWS) have an increased risk to develop tumors and receive tumor screening to monitor for tumor development. Research has shown that tumor risk varies depending on the molecular subtype of BWS. As a result, some groups began reassessing tumor screening recommendations and suggested screening only patients with molecular subtypes that are at higher risks. The effect of tumor screening on patients and parents has been argued as a reason to screen and not to screen. Benefits of tumor screening include early detection and giving patients/families a sense of control and continued reassurance.

Negative factors associated with tumor screening include increased anxiety, burden, and cost to patients and families. Although these factors exist, no study has evaluated patient and parental perspectives on the subject.

Purpose

This study aimed to identify how parents of children with BWS or isolated hemihypertrophy/hemihyperplasia (now called isolated lateralized overgrowth (ILO)) felt about tumor screening protocols. This was the first study that directly asked parents of children with BWS or ILO about their perspective on tumor screening.

Findings

A questionnaire was completed by 261 families with BWS/ILO from all over the world. Overall, the majority of families were worried that their child might develop a tumor. Most agreed that screening helped decrease their anxiety and disagreed that screening was a burden. Almost all families agreed that the potential of detecting a tumor early outweighed the anxiety around frequent screening. Similar responses were given by families from different countries, suggesting that despite the cultural differences, there is uniform awareness of the significance of tumor screening.

More than half (54.1%) of parents could not identify their child's tumor risk and more than one- fifth (23.3%) did not know their child's genetic cause. In regards to changing screening based on genetic type, 76.5% of parents did not think screening should vary by type. The majority of factors analyzed did not affect parental concern, with the exception of knowledge. Those with increased knowledge were more likely to believe in tumor screening according to genetic cause and be less worried if tumor screening recommendations were to change. Families living outside of the United States were more likely to correctly identify their child's tumor risk.

Conclusion

The discrepancy in knowledge between diagnosis and tumor risk suggests the need to better educate families about the decision-making process for screening. Tumor screening gives families a sense of control and reassurance about their child's health, and therefore is recommended for all BWS/ILO patients regardless of molecular cause.

Key Points

- Families should be educated about their specific molecular cause of BWS and associated tumor risk.
- Families are comforted by tumor screening and may experience additional stress if screening is discontinued.

Reference

Duffy KA et al. Tumor Screening in Beckwith-Wiedemann Syndrome: Parental Perspectives. *J Genet Couns.* 2017;27(4): 844-853. PubMed PMID: 29204812