# Clinical and Molecular Characteristics of Children with Beckwith-Wiedemann Syndrome and Isolated Hemi Hyperplasia at Sultan Qaboos University Hospital with Their Surveillance Outcomes

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## *Dear Editor,*

In a recent publication, Al-Hinai et al.[1] presented a pioneering effort to characterize Beckwith-Wiedemann spectrum (BWSp) disorders and associated tumor risk among Omani children. Their work is commendable for contributing valuable insights to the limited body of literature on BWSp in the Middle East.

The authors provide useful information about the clinical and molecular characteristics of nine children diagnosed with either Beckwith-Wiedemann Syndrome (BWS) or isolated hemihyperplasia (IHH). The incidence of macroglossia in BWS cases was 100%, while IHH cases consistently presented with lateralized overgrowth. These findings confirm phenotypic patterns of BWS and IHH reported globally while adding an important regional perspective.[2]

Tumor risk in BWSp varies greatly by molecular subtype, ranging from 1% to 30%.[3] Despite this well-known tendency, no embryonic malignancies or higher alpha-fetoprotein levels were recorded in this cohort. The authors attributed the results to the small sample size. Except for those with IC2 loss of methylation, the consensus guidelines recommend that serial abdominal ultrasonography tests continue until the age of seven.[2] This unanticipated finding raises exciting issues regarding putative protective modifiers, whether genetic, epigenetic, or related to prenatal and perinatal factors, that may be unique to this population and warrant further investigation.[3,4]

Furthermore, the study underscores a significant diagnostic challenge: all IHH cases tested negative for methylation abnormalities in blood, suggesting that molecular alterations may be limited to affected tissues and thus undetectable using blood-based assays. The authors emphasize the significance of developing more sophisticated diagnostic techniques, such as tissue-specific molecular testing, particularly in suspected mosaic instances.[5]

This study by Al-Hinai et al.[1] should be acclaimed for emphasizing the importance of ongoing surveillance protocols and providing baseline data on tumor risk in BWSp among children in Oman. Their work lays a foundation for future multicenter studies to explore genotype-phenotype correlations and to develop risk-adapted screening guidelines tailored to diverse populations.

## References

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