Oman Medical Journal [2026], Vol. 41, No. 3: e850

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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ARTICLE INFO

Article history:
Received: 1 May 2025
Accepted: 17 November 2025

Online:
DOI 10.5001/omj.2026.45

Dear Editor,

In the January 2025 issue of the *Oman Medical Journal*, Al-Hinai et al,[1] presented a pioneering effort to characterize Beckwith-Wiedemann spectrum (BWSp) disorders and the associated tumor risk among Omani children. Their study contributes valuable regional data to the limited literature on BWSp in the Middle East.

The authors describe the clinical and molecular characteristics of nine children diagnosed with either Beckwith-Wiedemann syndrome (BWS) or isolated hemihyperplasia (IHH). Macroglossia was present in all BWS cases, while all IHH cases presented with lateralized overgrowth. These findings are consistent with phenotypic patterns of BWS and IHH reported globally.[2]

Tumor risk in BWSp varies greatly by molecular subtype, ranging 1–30%.[3] Despite this well-known tendency, no embryonal malignancies or elevated alpha-fetoprotein levels were recorded in this cohort. The authors attributed this to the small sample size. Consensus guidelines recommend serial abdominal ultrasonography tests until seven years of age, except in patients with IC2 loss of methylation.[2] This unexpected finding raises important questions regarding possible protective modifiers, whether genetic, epigenetic, prenatal, or perinatal, 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 the molecular alterations may be confined to affected tissues and thus undetectable by blood-based assays. The authors emphasize the need for more sophisticated diagnostic techniques, such as tissue-specific molecular testing, particularly in suspected cases of mosaicism.[5]

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

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