# Updated tumor screening guidelines for Bohring-Opitz Syndrome

*New guidelines include screening for hepatoblastoma (liver cancer) in addition to existing Wilms tumor screening guidelines*

In April 2024, two additional individuals with Bohring-Opitz Syndrome (ASXL1-related disorder) were identified with hepatoblastoma[1], a rare pediatric cancer of the liver. This is in addition to another case first reported in April 2023[2].

Hepatoblastoma is an extremely rare pediatric cancer (1 in a million in the general population). With 3 known instances of hepatoblastoma in an estimated 300 individuals with Bohring-Opitz Syndrome, **the risk of hepatoblastoma in individuals with Bohring-Opitz Syndrome remains very low (less than 1%), but it is significant enough to warrant new screening recommendations.**

Hepatoblastoma typically affects young children in the first 5 years of life. **Individuals with hepatoblastoma are typically cured if their cancer is identified and treated early.** Routine screening can be done via abdominal ultrasound to look for tumors developing in the liver, and by a blood test to measure level of alpha-fetoprotein (AFP), a protein made by the liver.

These new screening recommendations for hepatoblastoma are *in addition to* the existing screening recommendations for Wilms tumor[3], a rare pediatric cancer of the kidneys, which include a screening ultrasound to look for tumors developing on the kidneys every three months until age 8.

**Updated tumor screening recommendations include:**

- Abdominal ultrasound *including imaging of the kidneys and liver* every three months until age 8
- Discussion of the following additional screening options with your child's care team:
    - Measurement of alpha-fetoprotein (AFP) via blood draw every three months until age 5
    - Continuation of ultrasound screening past age 8 at a reduced frequency (e.g. every 6 months)

These recommendations are made by the ARRE Foundation Medical and Scientific Advisory Board, including Dr. Bianca Russell (University of California, Los Angeles) and Dr. Wen-Hann Tan (Boston Children's Hospital) who are clinical geneticists with longstanding clinical research interests in Bohring-Opitz Syndrome.

For more information about Bohring-Opitz Syndrome, including links to relevant clinical research publications, please visit arrefoundation.org/bohring-opitz

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**References:**

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2: Russell, B. E., Kianmahd, R. R., Munster, C., Yu, A., Ahad, L., & Tan, W. H. (2023). Clinical findings in 39 individuals with Bohring-Opitz syndrome from a global patient-driven registry with implications for tumor surveillance and recurrence risk. *American journal of medical genetics.* Part A, 191(4), 1050–1058. https://doi.org/10.1002/ajmg.a.63125

3: Russell, B., Johnston, J. J., Biesecker, L. G., Kramer, N., Pickart, A., Rhead, W., Tan, W. H., Brownstein, C. A., Kate Clarkson, L., Dobson, A., Rosenberg, A. Z., Vergano, S. A., Helm, B. M., Harrison, R. E., & Graham, J. M., Jr (2015). Clinical management of patients with ASXL1 mutations and Bohring-Opitz syndrome, emphasizing the need for Wilms tumor surveillance. *American journal of medical genetics. Part A*, *167A*(9), 2122–2131. https://doi.org/10.1002/ajmg.a.37131