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## PAPER OF THE MONTH • DECEMBER 2019



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## Mesenchymal Hamartoma of the Liver and *DICER1* Syndrome

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For a decade, Dr. William Foulkes and his collaborators have been puzzling over various mutations and alterations of the gene *DICER1* in order to more fully comprehend its catalytic activity. In this report, they examine the only two documented cases so far identified of *DICER1* mutations in mesenchymal hamartoma of the liver (MHL), the second most common benign liver tumor in children. Prior to this discovery, there was nothing to suggest that this gene was implicated in MHL. While two cases are not definitive, they do offer a new clue as to the effects *DICER1* mutations may have.

This research is also noteworthy for employing refined biochemical techniques that improve the capacity to predict the functional effect of specific variants within the gene. Work done by Dr. Maria Apellaniz-Ruiz, a post-doctoral fellow, working in Dr. Foulkes' lab at the LDI, in a close collaboration with Dr. Marc Fabian, contributed the complex sequencing analysis. The work is indicative of the collaborative efforts between Dr. Foulkes, working on the clinical and molecular genetics side, and Dr. Fabian, on the biochemical. The paper also represents a far-ranging collaboration between the LDI and clinicians in Italy and Germany, where the two children were treated.

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