In the news

PROBING A RARE TUMOUR TYPE

A recent *Science* paper reported a recurrent chimeric transcript in fibrolamellar hepatocellular carcinoma (FL-HCC), a rare liver tumour that affects adolescents and young adults (*Science* **343**, 1010–1014; 2014). Interestingly, two of the authors on the paper are survivors of FL-HCC: one asked not to be identified, but the other, high school student Elana Simon, was substantially involved in the study and was a co-first author.

One of the biggest challenges in studying FL-HCC is the lack of samples, as only about 200 cases are diagnosed each year worldwide. The authors were able to obtain 15 tumour samples, in part by Simon reaching out to patient groups. In all of the 15 samples, they found a deletion on chromosome 19 that resulted in the fusion of the amino terminus of DNAJB1 (a homolog of the DNAJ chaperone protein) to the carboxyl terminus of the protein kinase A catalytic domain (PRKACA), and this fusion protein retains kinase activity. Sanford Simon (Rockefeller University, New York, USA), the corresponding author of the paper (and Elana Simon's father) noted that "It is uncommon for a genetic screen for a cancer to turn up such a strong candidate mutation, and for the mutation to be present in every single patient tested" (*PR Newswire*, 28 Feb 2014).

Elana Simon had used her background in computer science to examine mutations in a different tumour type during an internship in another lab, and she thought that this approach might help to understand FL-HCC. Given how much research can now be done using computers, her father reasoned that this is "an exciting time for kids to go into science" (*Associated Press*, 27 Feb 2014).

This study highlights the impact survivors can have on research. As Elana Simon stated, "someone had to do something about this disease, and since nobody else was going to, I decided to take it upon myself" (*PR Newswire*, 28 Feb 2014).

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