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DGCR8 microprocessor defect characterizes familial multinodular goiter with schwannomatosis

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Upon being introduced to a Montreal family with a strange confluence of multinodular goiter (MNG) and schwannomatosis across three generations, a team led by Dr. William Foulkes investigated and discovered a brand-new syndrome brought about by a rare genetic mutation. The finding is interesting because it shines a light on unknown biology. The link between inherited multinodular goiter and the development of schwannomatosis had never been observed before. Having access to three generations with the same unusual conditions, the researchers had the opportunity to conduct whole-exome sequencing in order to find out whether they could isolate the causal mutation.

Following painstaking analysis, the gene known as “Di George Critical Region 8” (DGCR8) was identified as the culprit in what the authors of the paper dubbed *Familial MNG with schwannomatosis*. A single mutation that slightly alters the protein results in deficient production of microRNAs, bringing about this rare syndrome. This study represents an impressive scientific exercise, where a dedicated group of scientists from around the world worked to solve a problem that the team at the JGH were the first to recognize as a new syndrome.

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