This week in techniques

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Studies in patients and mice suggest a mutation in NLRC4 could be a diagnostic marker for FCAS, which involves episodes of rash, arthralgia and fever after exposure to cold stimuli. Genomic sequencing of seven non-consanguineous members of a Japanese family identified a heterozygous 1589A>C missense mutation in NLRC4 in the five individuals who had the condition but not in the two individuals who did not. In mice, the equivalent missense mutation in Nlrc4 yielded animals that recapitulated symptoms of human FCAS, such as systemic autoinflammation in response to cold stimuli. Next steps could include validating the missense mutation as a marker of FCAS in larger patient cohorts.

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