



## Response to: 'Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype' by Gupta *et al*

We read with great interest the letter by Aggarwal.<sup>1</sup> Her case is one of early-onset autoinflammatory disease.

Her case is not directly similar to any of the 187 cases we described in our paper on undefined autoinflammatory diseases.<sup>2</sup>

Given the finding of novel potentially pathogenic NLRP12-variants in homozygosity, we think the authors have most likely identified a hitherto unknown cause of this child's symptoms. As such, we recommend the case with all its details to be submitted for publication in a scientific journal.

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### REFERENCES

- 1 Gupta L, Ahmed S, Singh B. Novel NLRP12 variant presenting with familial cold autoimmunity syndrome phenotype. *Ann Rheum Dis* 2021;**80**:e117.
- 2 Ter Haar NM, Eijkelboom C, Cantarini L, *et al*. Clinical characteristics and genetic analyses of 187 patients with undefined autoinflammatory diseases. *Ann Rheum Dis* 2019;**78**:1405–11.