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Germline biallelic PIK3CG mutations in a multifaceted
immunodeficiency with immune dysregulation

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Contributions: MT conceptualized, designed, and performed experiments, analyzed data, and wrote the manuscript. BH performed and analyzed neutrophil assays and edited the manuscript. AK performed macrophage morphology assays and image analysis. FP, CH and AA took care of the patient, provided patient samples and interpreted clinical data. SKB and JH provided experimental help. MC analyzed macrophage morphology data. RJH conducted and analyzed NGS data, and identified PIK3CG mutations in the patient. WFP performed immunophenotyping and analysis. MG established and performed initial NK-cell degranulation assays. SE, CLL, JM, and LD gave important intellectual input. KB supervised the study, reviewed clinical and experimental data, and wrote the manuscript. All authors reviewed and approved the manuscript.