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Executive Summary

The overall aim of WP6 is to identify the genetic basis of MIS-C/A and COVID toes. This deliverable addresses the dissemination of results from this work package.

Abbreviations

D	Deliverable
EC	European Commission
WP	Work Package
WT	Work Task

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1 Dissemination for the scientific and medical community

Academic courses, and meetings related to UNDINE:

- Danyel Lee of the HGID lab (INSERM) presented the studies of new candidate MIS-C causing genes at the COVID Human genetics effort monthly meeting in March.

Conferences in which UNDINE-results were presented:

- Vanessa Sancho Shimizu presented the genetic basis of MIS-C at the Disease Models and Mechanisms: Infectious Diseases Through an Evolutionary Lens meeting in London, UK October 17-19 2023
- Vanessa Sancho Shimizu presented the role of BTNL8 in MIS-C at the fall symposium of the Section for Paediatric Infectious Diseases and Immunology of the Dutch Paediatric Society November 3, 2023
- Vanessa Sancho Shimizu presented the genetic basis of MIS-C at the IUIS meeting in Capetown South Africa December 2, 2024
- Vanessa Sancho Shimizu presented *BTLN8* variants in MIS-C: lessons for KD pathology? 2nd IKDR and EUROKiDs Joint Kawasaki Conference, Amsterdam March 4-6, 2026
- Shen-Ying Zhang presented gain-of-activity of the RIG-I pathway in MIS-C at the 20th International Symposium on Inborn Errors of Immunity, Newport Beach, CA, USA

Overview of publication efforts with open access:

- Inborn errors of OAS-RNase L in SARS-CoV-2-related multisystem inflammatory syndrome in children. Lee D, Le Pen J, Yatim A ... Meyts I ... Pujol A, Sancho-Shimizu V ... Perez de Diego R ... Abel L ... Quintana-Murci L ... Zhang SY, Casanova JL. *Science*. 2023 Feb 10; 379(6632):eabo3627. doi: 10.1126/science.abo3627. Epub 2023 Feb 10. PMID: 36538032
- Rare predicted loss-of-function variants of type I IFN immunity genes are associated with life-threatening COVID-19. Matuozzo D, Talouarn E, Marchal A ... Casari G ... Martinez-Picado J ... Novelli A ... de Diego RP ... Pujol A ... Soler-Palacin P ... Fellay J ... Mogensen TH ... Meyts I, Zhang SY ... Casanova JL ... Abel L, Cobat A. *Genome Med*. 2023 Apr 5; 15(1):22. doi: 10.1186/s13073-023-01173-8. PMID: 37020259; PMCID: PMC10074346
- Autoantibodies Neutralizing Type III Interferons Are Uncommon in Patients with Severe Coronavirus Disease 2019 Pneumonia. Vanker M, Särekannu K, Fekkar A, ... Zhang SY, Mogensen TH, ... Casanova JL, Kisand K. *J Interferon Cytokine Res*. 2023 May 29; doi: 10.1089/jir.2023.0003. Epub ahead of print. PMID: 37253131. PMID: 37253131

- Bellos E, Santillo D, Vantourout P, Jackson HR, Duret A, Hearn H, Seeleuthner Y, Talouarn E, Hodeib S, Patel H, Powell O, Yeoh S, Mustafa S, Habgood-Coote D, Nichols S, Estramiana Elorrieta L, D'Souza G, Wright VJ, Estrada-Rivadeneira D, Tremoulet AH, Dummer KB, Netea SA, Condino-Neto A, Lau YL, Núñez Cuadros E, Toubiana J, Holanda Pena M, Rieux-Laucat F, Luyt CE, Haerynck F, Mège JL, Chakravorty S, Haddad E, Morin MP, Metin Akcan Ö, Keles S, Emiroglu M, Alkan G, Tüter Öz SK, Elmas Bozdemir S, Morelle G, Volokha A, Kendir-Demirkol Y, Sözeri B, Coskuner T, Yahsi A, Gulhan B, Kanik-Yukse S, Bayhan GI, Ozkaya-Parlakay A, Yesilbas O, Hatipoglu N, Ozcelik T, Belot A, Chopin E, Barlogis V, Sevketoglu E, Menentoglu E, Gayretli Aydin ZG, Bloomfield M, AlKhater SA, Cyrus C, Stepanovskiy Y, Bondarenko A, Öz FN, Polat M, Fremuth J, Lebl J, Geraldo A, Jouanguy E; COVID-19 Human Genetic Effort; DIAMONDS; EUCLIDS; Carter MJ, Wellman P, Peters M, Pérez de Diego R, Edwards LA, Chiu C, Noursadeghi M, Bolze A, Shimizu C, Kaforou M, Hamilton MS, Herberg JA, Schmitt EG, Rodriguez-Palmero A, Pujol A, Kim J, Cobat A, Abel L, Zhang SY, Casanova JL, Kuijpers TW, Burns JC, Levin M, Hayday AC, Sancho-Shimizu V. Heterozygous BTNL8 variants in individuals with multisystem inflammatory syndrome in children (MIS-C). *J Exp Med*. 2024 Dec 2;221(12):e20240699. doi: 10.1084/jem.20240699. Epub 2024 Nov 22. PMID: 39576310; PMCID: PMC11586762.
- Santillo D, Bellos E, Sancho-Shimizu V. Unravelling $\gamma\delta$ T-cell dysregulation in the gut and its implications for immune-mediated diseases. *Dis Model Mech*. 2025 Sep 1;18(9):dmm052439. doi: 10.1242/dmm.052439. Epub 2025 Sep 18. PMID: 40965151; PMCID: PMC12486229.
- Lee D, Malathi K, Okano T, Nakajima K, Cobat A, Morio T, Casanova JL, Zhang SY. The OAS-RNase L pathway: insights from experiments of nature. *Science Immunology*, 2026; 11(115):eads9407.