

Rare 2030 Panel of Experts

Membership List for Sub-Group 5: Diagnostics*

Surname	First Name	Type of Stakeholder**	Country
Aleksovska	Vesna	Patient advocate	MK
Alves	Inês	Patient advocate	PT
Amin	Ahmad	ERN Coordinator/ERN Healthcare Provider representative	NL
Anastasiadou	Violetta	National/Regional Competent Authority (National policy maker)	CY
Barić	Ivo	National/Regional Competent Authority (National policy maker)	HR
Baynam	Gareth	International initiatives	AT
Benfredj	Rudy	Pharmaceutical Industry and other health-related industries	UK
Bols	Thomas	Pharmaceutical Industry and other health-related industries	CH
Bros-Facer	Virginie	European research networks and infrastructures	FR
Bruijns	Christel	Pharmaceutical Industry and other health-related industries	CH
Buske	Orion	Individual Researcher	CA
Dallapiccola	Bruno	National/Regional Competent Authority (National policy maker)	IT
D'Amato Sizonenko	Loredana	Patient advocate	CH
Digneffe	Toon	Pharmaceutical Industry and other health-related industries	BE
Dooms	Marc	Individual Researcher	BE
Evangelista	Teresinha	ERN Coordinator/ERN Healthcare Provider representative	UK
Evers	Pauline	Regulatory Expert	NL
Graessner	Holm	ERN Coordinator/ERN Healthcare Provider representative	DE

Ingeborg	Barisic	Individual expert involved in the European Joint Actions for Rare Diseases	HR
Isla Gomez	Julian	Regulatory Expert	ES
Jacinto	Marta	Patient advocate	PT
Jondeau	Guillaume	ERN Coordinator/ERN Healthcare Provider representative	FR
Kaariainen	Helena	National/Regional Competent Authority (National policy maker)	FI
Konecna	Katerina	EU Policy Maker	CZ
Lohse	Ansgar	ERN Coordinator/ERN Healthcare Provider representative	DE
Lopez Granados	Eduardo	ERN Coordinator/ERN Healthcare Provider representative	ES
Macek	Milan	National/Regional Competent Authority (National policy maker)	CZ
Malniece	Leva	Individual Researcher	LV
Meade	Nick	Patient advocate	UK
Melegh	Bela	Individual experts involved in the European Joint Actions for Rare Diseases	HU
Miasnikova	Irina	Patient advocate	RU
Muthyala	Ramaiah	International initiatives	US
Nabbout	Rima	European research networks and infrastructures	FR
Oosterwijk	Cor	Patient advocate	NL
Özbek	Uğur	Individual Researcher	TR
Palau	Francesc	Individual experts involved in the European Joint Actions for Rare Diseases	ES
Pirard	Vinciane	Pharmaceutical Industry and other health-related industries	BE
Posada	Manuel	Individual experts involved in the European Joint Actions for Rare Diseases	ES
Rui Santos	Ivo	Regulatory Expert	PT
Scarpa	Maurizio	ERN Coordinator/ERN Healthcare Provider representative	IT
Schoeters	Eva	Patient advocate	BE
Severin	Emilia	National/Regional Competent Authority (National policy maker)	RO

Spink	Jayne	Patient advocate	UK
Taruscio	Domenica	National/Regional Competent Authority (National policy maker)	IT
Treacy	Eileen	National/Regional Competent Authority (National policy maker)	IE
Ussi	Anton	European research networks and infrastructures	NL
van Doorne	Cathalijne	Patient advocate	BE
van Zelst-Stams	Wendy	National/Regional Competent Authority (National policy maker)	NL
Verloes	Alain	ERN Coordinator/ERN Healthcare Provider representative	FR
Wong-Rieger	Durhane	International initiatives	CA
Zain	Rula	Individual Researcher	SE
Zerjav Tanšek	Mojca	National/Regional Competent Authority (National policy maker)	SI

(NB The following panel members were *assigned* to this group and may wish to change!)

Surname	First Name	Type of Stakeholder**	Country
Daly	Avril	Patient advocate	IE
Jondeau	Guillaume	ERN Coordinator/ERN Healthcare Provider representative	FR
Miasnikova	Irina	Patient advocate	RV
Valdmane	Antra	National/Regional Competent Authority (National policy maker)	LV
Zhang	Shuyang	International Initiatives	CN

*This subgroups includes topics such as Prevention, Newborn screening, Next-Generation Sequencing & its introduction to the clinic; AI; undiagnosed patients; genetic counselling

**Many people in this Panel of Experts belong to numerous categories – the ‘Type of Stakeholder’ listed against each name represents the *primary* capacity in which experts were invited to this Panel.