



TOOL 6

Brief Summaries of the first Together4RD Pilots



Osteogenesis Imperfecta, natural history and innovative clinical trial measures

This pilot project centres of three main activities:

Firstly, value will be drawn from several sources of existing data relating to patients with the rare bone

condition OI, to better elucidate the natural history and the disease burden. Genotype-phenotype correlations will be explored, as part of this drive to better understand the condition.

Secondly, prospectively, the project will measure the impact of disease on patient activity and quality of life using digital technologies in a real-world setting, through a combined approach of gait analysis and sensors.

Lastly, in preparation for smoother regulatory pathways for OI therapies in future, the project partners will jointly engage in early scientific dialogues with other stakeholders (such as regulators and HTA), to gain insights into approaches to foster patient access to innovative medicines.



Improving time to diagnosis for unmet patient needs in rare hematologic diseases

Who is involved? EuroBloodNet, Innova, ENROL and Takeda in partnership

This initiative is all about improving the diagnostic pathways for rare haematological diseases by

utilizing AI algorithms for early detection. The activity is expected to focus on thrombotic thrombocytopenic purpura (TTP), a very rare and debilitating condition existing in both a congenital form and an acquired, immune-mediated form (which manifests in the 4th or 5th decades). The variable phenotype for TTP and lack of availability of the necessary diagnostic tests, leads to delayed diagnosis, which is very concerning as without a correct diagnosis and

treatment, organ ischemia and death occurs in around 90% of cases.

To improve early diagnosis, the plan is to employ an AI federated platform across key clinical centres, which will ideally be based on the hospitals own EHR systems. Whilst reducing time to diagnosis, and thus enabling treatment to begin as early as possible, the AI platform should lead directly to improved patient outcomes by alerting treating clinicians of patients at risk of relapse, who can then be monitored in appropriate ways.

The initial phase of the pilot will focus on scoping activities and identifying centres for the implementation phase. From the ERN side, the pilot is being delivered through an organisation known as the EuroBlooNet Association, together with the Fundació Hospital Universitari Vall D'Hebron, which is the entity in charge of the ERN's shared registry (ENROL), and also the Fondazione IRCCS Ca' Granda - Ospedale Maggiore Policlinico.



RHINE Project: Rare Renal and PH International Network

Who is involved? Novo Nordisk, ERKnet and OxalEurope

The RHINE Project aims to improve care pathways and patient outcomes in rare renal conditions, by building on existing registry infrastructure, especially the core registry of the ERK-Net ERN (named **ERK-Reg**). This

pilot focuses in particular on an ultra-rare disease called primary hyperoxaluria (PH), and addresses a fundamental need to complement the core registry structure of the ERN with detailed data collection for specific conditions.

A 'harmonization and interoperability' model will be developed for PH, by establishing a Rare Kidney Network data registry, in collaboration with the European Hyperoxaluria Consortium (OxalEurope), to achieve seamless data connection across ERK-Net and PH-specific registries.

Through this new data linkage approach, the project aims to understand PH diagnostic and referral pathways locally, measuring metrics like time-to-diagnosis, where PH patients are seen (by measuring the number of cases in each referring center), and the percentage of patients diagnosed before they reach end-stage renal disease (the point at which the kidneys can no longer support the body's needs). To compliment this focus on an improved data ecosystem, educational activities will be implemented at regional and local levels, to

target the gaps and shortcomings identified. As of late 2024, this pilot has already generated significant added- value simply by bringing the registry owners from the ERK-REG and the OxalEurope consortia together, around the same table, having built a robust formal agreement for collaboration. The technical integration work is now underway, mapping data dictionaries to explore what each registry ecosystem currently collects.



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