ERDERA
European Rare Diseases
Research Alliance

Rare and Genetic Diseases Specificities: Case Studies

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Persona Case Study
Name: Emma Davis
Age: 13
Diagnosis: Phenylketonuria (PKU)
Diagnosis Age: Diagnosed at 5 days old through the national newborn screening programme
Location: Manchester, UK
Family Background: Lives with both parents and a younger brother (age 9)
School: Attends a mainstream school

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Persona Case Study:

Name: Nicholas O'Reilly
Age: 4
Diagnosis: Metachromatic leukodystrophy (MLD) is a genetic disorder that affects nerves, muscles, other organs, and behaviour. It slowly gets worse over time.

Diagnosis Age: Diagnosed at 18 months of age

Location: Dublin, Ireland

Family Background: Lives with both parents and an older sister (age 8)

Gene therapy clinical trial in Italy

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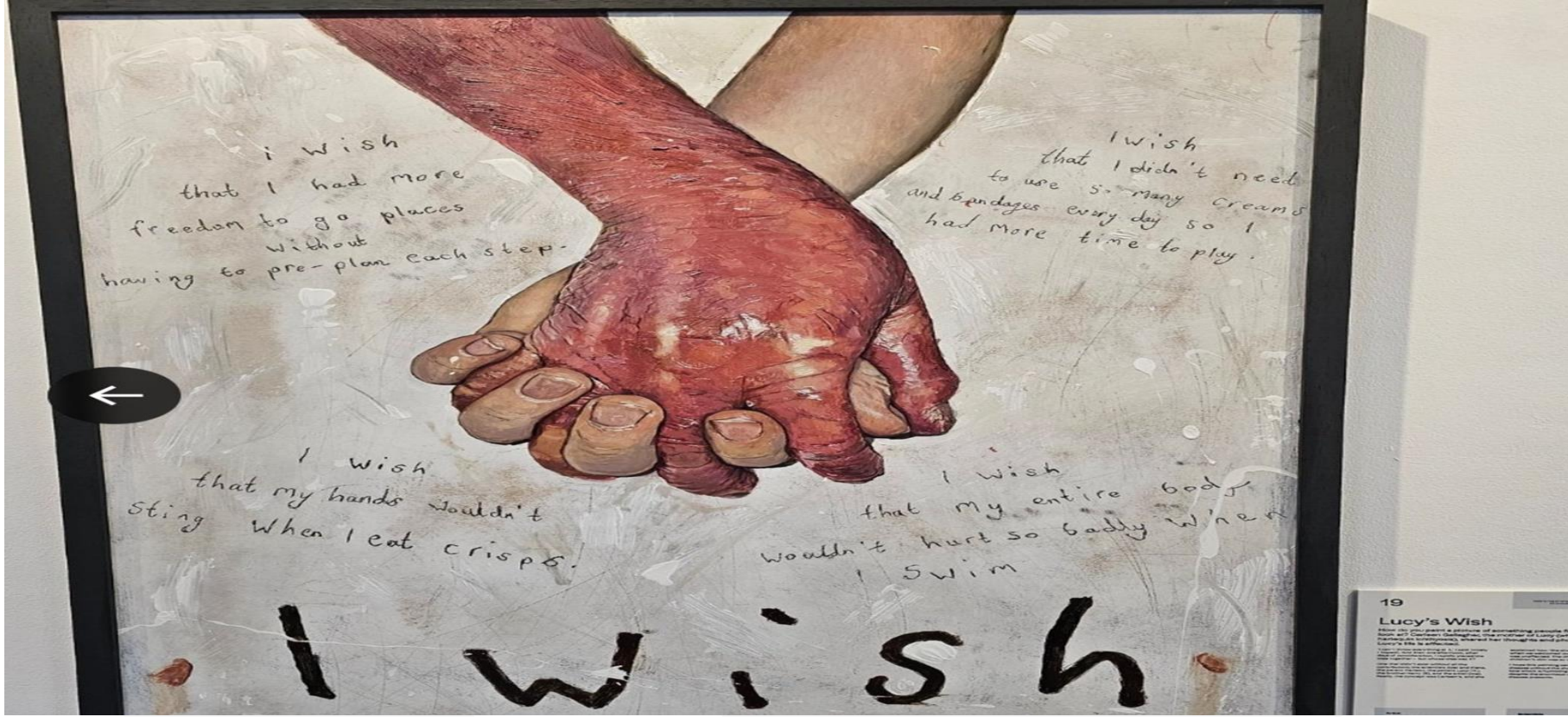

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Making Europe a world-leader in rare diseases research and innovation

Connecting the dots in the rare diseases research ecosystem

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