European Reference Network for Rare Neurological Diseases
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Le Tribunal Judiciaire de Lyon
Pôle Social, Contentieux Général
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France

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Subject: Case N° RG 19/00047, N° Portalis DB2H-W-B7D-TQUU (CG), amicus curiae brief

Dear Tribunal Judiciaire de Lyon,

I am writing as an amicus curiae to provide insight and expertise in the case involving, as plaintiffs, Lysiane Pakter and her mother Delphine Beulné, and as defendant, La Caisse Primaire d'Assurance Maladie (CPAM) du Rhône, Case N° RG 19/00047, N° Portalis DB2H-W-B7D-TQUU (CG).

I will begin by providing information on my academic qualifications and my professional experience, which focuses on rare diseases. I will then identify the legal issues which I consider to be central to the case, and I will share my views on those issues. After addressing the principal legal issues, I will analyse the case from a public policy perspective. This will be followed by a conclusion which summarizes the points I have raised.

Academic qualifications and professional activities

I carried out my studies in Biomedical Engineering, Cybernetics, Electrical Engineering, German Language and Literature, Philosophy, and Business Administration. In 2004 I received my PhD, *summa cum laude*, and in 2008 I obtained an MBA degree.

I have been working in rare diseases for over two decades now. I am both a healthcare manager and a research manager. As a healthcare manager I am the Managing Director of the Centre for Rare Diseases at the Tübingen University Hospital in Tübingen, Germany, since 2010 and the Coordinator of the European Union's European Reference Network for Rare Neurological Diseases (ERN-RND) since 2017. As a research manager, my team and I have been coordinating more than 30 large global, European and national collaborative research projects focusing on rare diseases. I will provide some details below.

I have been deeply involved with the European Union's European Reference Networks (ERNs) since their inception. In 2017 I launched the European Reference Network for Rare Neurological Diseases (ERN-RND), and was appointed as its Coordinator. ERN-RND is made up of the European clinical expertise centres which have been carefully selected and are experts in the different rare neurological

disease groups. We work together to improve the diagnosis and treatment of rare neurological diseases.

I am also involved with the ERN structure on a higher level, in the ERN Coordinators Group, where I served as the co-chair of this group.

In 2018 I began serving as the Coordinator of Solve-RD, a flagship project funded by the European Commission. Solve-RD works with the ERNs, carries out research in order to diagnostically solve rare diseases for which a molecular cause is not yet known.

Together with Allianz Chronischer Seltener Erkrankungen (ACHSE), Germany's national rare disease patient organization, I organised the first National Rare Disease Conference in Germany, in 2019, and again in 2021 and 2023.

Last year, in 2023, the European Organisation for Rare Diseases (EURORDIS) honoured me with the EURORDIS Black Pearl Award for Leadership, in recognition of "lifelong dedication to the rare disease community and the coordination of countless projects at a national, European and international level".

The legal issues which are central to the case

The following are excerpts from the draft request for a preliminary ruling, dated 23 January 2024. These excerpts from the draft request for a preliminary ruling bring us to one of the core legal issues of the case, and therefore I will present them below:

> 34. As reported in ***The Lancet***, there are over 6,000 rare diseases that we know of today; 95% of them have no approved treatment or cure. Half of these rare diseases strike children (Nguengang et al. have shown that 70% of the diseases that Orphanet contains have an exclusively pediatric onset); 30% of these children will die before they are five years old.
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> 35. For approximately 5% of rare diseases, there are effective treatments or cures. These highly specialized rare disease treatments are often only available in certain geographically limited locations, at an officially designated Orphanet Centre of Expertise, which focuses on treating patients with that particular rare disease.
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> 36. Regulation 883/2004 can make a profound difference for a rare disease patient, by allowing them to access a highly specialized rare disease treatment which is not available where that patient lives, but which is available elsewhere in Europe.
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> 42. Thus: rare disease patients have a particularly strong need for planned cross-border healthcare, in order to access highly specialized treatments which are not available where they live – and the total cost associated with planned cross-border healthcare for rare disease patients amounts to an extremely small portion of total national healthcare spending in Europe.
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> 45. Lysiane's parents argue that based on exclusively medical criteria – the fact that rare disease patients face grave health problems, have very limited treatment options, and are often very young and vulnerable children – a rare disease patient's S2 application should not be processed in the same way as an S2 application for dental care or eyeglasses or other routine healthcare services. They claim that based on the Charter's principle of equal treatment and non-discrimination, which is enshrined in Article 20 and 21(1), a Member State should consider a broader range of scientific evidence, and provide a more extensive justification, and meet a higher threshold of necessity and proportionality, when refusing to

> grant an S2 to a rare disease patient seeking access to a safe and effective treatment which is only available in another Member State of the European Union.

Thus, one of the core questions we must address is the following: does European Union law oblige public authorities and healthcare insurers to apply special measures when a rare disease patient submits an S2 application to access a safe and effective highly specialised treatment which is not available where they live, but which is available elsewhere in Europe?

In response to this core legal question, Dr. Barend van Leeuwen, Professor of European Union Law, points out in his amicus curiae legal brief dated 8 May 2024 that patients with rare diseases "are inherently more vulnerable as a patient group, because of the lack of expertise available to diagnose and treat their diseases". He also notes that in order to access a highly specialized rare disease treatment, rare disease patients are "significantly more likely to require cross-border healthcare than patients with more regular illnesses or diseases".

Based on these facts, and in accordance with Article 20 and 21 of the Charter of Fundamental Rights of the European Union, which guarantee equality and non-discrimination, rare disease patients do have the right to additional legal protection when seeking access to evidence-based treatments which are not available where they live, but which are available elsewhere in Europe. I agree with Professor van Leeuwen's legal position: "more onerous and more precise obligations should be imposed on public authorities or health insurers when they are dealing with requests for prior authorisation submitted by patients with rare diseases".

Based on my professional experience, I share Professor van Leeuwen's view, and I join him in encouraging the court to make a preliminary reference to the CJEU, in order to obtain definitive answers to these important legal questions.

Public policy implications

In addition to the important legal questions which are raised by this case, there are also significant public policy issues involved.

As a matter of public policy, we can all agree on the following fundamental and universal principle: access to healthcare should be based on medical needs, not on financial means.

This fundamental, universal principle, that access to healthcare should be based on medical needs, and not on financial means, is especially important when we are dealing with a highly vulnerable patient suffering from a complex rare disease. Rare disease patients and their families already carry a very heavy burden; public policy frameworks should therefore do everything reasonably possible to ease this burden, not add to it.

The UN's Sustainable Development Goals call upon all nations of the world to "leave no one behind". This global commitment is especially relevant for rare disease patients, as highlighted in the draft request for a preliminary ruling, dated 23 January 2024:

> 43. The 2021 UN Resolution, "Addressing the challenges of persons living with a rare disease", calls upon nations to eliminate gaps in coverage for rare disease patients. The UN 2030 Agenda's Sustainable Development Goals specifically identify rare disease patients as a vulnerable patient group, which merits special assistance. "The 2030 Agenda places emphasis on equality, and the commitments to leave no one behind, and to reach first those who are furthest behind, are founded on the human rights principles of equality and non-discrimination, and of prioritizing the vulnerable and marginalized in society... States should ensure, in particular, that the legal and policy framework addresses discrimination in access

> to health care and services, ensures effective access to medicines, therapies and technologies for all persons without discrimination, and protects the rights of persons living with rare disease… and other vulnerable groups, including through the use of special measures where appropriate".

Rare disease patients face many difficult challenges, including high morbidity, high mortality, an enormous scarcity of safe and effective treatments – and rare diseases disproportionately strike children. Rare disease patients are certainly "behind", and the UN High Commissioner for Human Rights is correct: the human rights principles of equality and non-discrimination justify special public policies to ensure that highly vulnerable rare disease patients can actually access the medicines, therapies and technologies they need.

Conclusion

Rare disease patients require additional legal protection, and public authorities and health insurers require greater legal clarity, both of which the CJEU can provide through a preliminary ruling. For both legal and public policy reasons, and consistent with the fundamental principles of equality and non-discrimination, I encourage the court to make a preliminary reference to the CJEU.

Best regards,

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