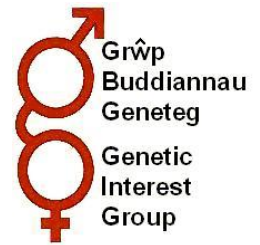


# Consultation Response

Inquiry into the proposed EU Directive on Patients' Rights in Cross Border Healthcare

National Assembly for Wales European and External Affairs Committee

Response by the Genetic Interest Group



## Introduction

The Genetic Interest Group (GIG) is the UK national alliance of families and all those affected by genetic conditions. GIG works to raise awareness and improve the health service provision available to those living with and at risk from inherited conditions. Our membership represents 138 voluntary organisations working for a wide range of conditions, many of which are rare. GIG welcomes the National Assembly for Wales' European and External Affairs Committee's inquiry into the proposed EU Directive on Patients' Rights in Cross Border Healthcare.

In GIG's view, NHS Wales as a separate body to the NHS in England, governed by the Welsh Assembly Government, must communicate and work with the Department of Health to ensure its views are represented on a European level, ensuring patients and their families in Wales benefit as much as possible from the opportunities cross border healthcare may offer.

Our response to this consultation focuses on the needs of our member organisations and the individuals and families that they represent.

## The special case of rare genetic disease

More than 6,000 genetic conditions are currently identified, and most are rare (falling into the EU definition of a prevalence lower than 5 in 10,000). Understandably, due to this low prevalence, levels of awareness for many conditions will be poor. Most rare conditions will only be seen by a particular health professional once in their career, or not at all, and so it is impossible to develop the clinical expertise to correctly diagnose and implement the best possible management of care and treatment for every rare disease in all areas of Wales.

Rare disease itself is not rare though; more than 175,000 people living in Wales will be affected by a rare disease at some point in their lives. Clinicians should therefore be aware of the likelihood of being presented with a patient with a rare genetic disease, and should be able to access expertise wherever it may be. Borders within the UK or within the EU should not be barriers to proper exchange of this valuable knowledge and expertise.

Genetic conditions are life-long and / or multifactorial. Onset times vary between conditions and sometimes within the same condition, from childhood to old age; symptoms may also differ in two patients with the same diagnosis (phenotypic variability). Most genetic conditions are incurable and many are without treatment. Conditions require specialist management of care and diagnosis to ensure those affected receive the best and most appropriate service, often from multiple disciplines.

This requirement for specialist care, coupled with the rarity of most genetic conditions points to the great benefits that healthcare for genetic conditions can gain from sharing resources across borders within both the UK and the EU. Centres of excellence need to have a large enough population of patients to be cost effective and maintain high standards of care and safety; for some very rare conditions, the population of Wales, and indeed the population of the UK, is not large enough to sustain them.

## **Other potential benefits for Wales from cross border healthcare**

Aside from patients crossing borders, there are other opportunities to improve healthcare delivery arising from this proposed directive. The principle of cooperation between countries and regions should also be applied to information such as Health Technology Assessments and standards of care from areas of established excellence in other Member States.

Stronger networking across Europe could also benefit innovation by enhancing research and development programmes, as well as establishing international clinical trials for new therapies and treatments or for developing registries for particular conditions. This is of particular importance for conditions which are very rare, studies are often not feasible due to small patient cohorts for research programmes in Wales or the UK.

## **Existing European Cross Border initiatives benefiting healthcare**

Clinicians with an interest in medical specialties are already working with colleagues across Europe to improve knowledge that will provide better healthcare for their patients. Clinical geneticists based at the Institute of Medical Genetics are involved in the TREAT-NMD project – an European Union funded network of expertise that involves 21 partner organisations from 11 EU Countries (including the UK). Since its establishment in 2007, it has encouraged harmonisation and reduced fragmentation in the neuromuscular community, working to accelerate clinical trials and also promote common standards of care for those with muscle diseases such as muscular dystrophy.

Another initiative is the European Network for the Study of Orphan Nephropathies (kidney disease), established in order to provide the required critical mass of expertise on a European-wide scale for the study of orphan nephropathies with patient registries and a network of medical genetics laboratories.

This Directive is an opportunity to strengthen existing networks and ensure that more clinicians working in Wales can access the best possible knowledge, information and resources so that they can give a high level of care to their patients.

GIG welcomes the National Assembly for Wales European and External Affairs Committee's inquiry into this directive, and will be happy to comment further.