Dearadh agus Nuálaíocht Chliniciúil, Oifig an Príohoifigeach Cliniciúil
Ospidéal Dr. Steevens, D08 W2A8
R: clinicaldesign@hse.ie

Clinical Design & Innovation, Office of the Chief Clinical Officer
Dr Steevens' Hospital, D08 W2A8
E: clinicaldesign@hse.ie

22nd April, 2025

Deputy Pádraig O'Sullivan, TD
Dáil Éireann
Leinster House
Kildare Street
Dublin 2

PQ 16457/25
To ask the Minister for Health her plans to ensure timely access to diagnostics and treatment for rare diseases such as transthyretin amyloid cardiomyopathy (ATTR-CM); and if she will make a statement on the matter

Dear Deputy O'Sullivan,

The Health Service Executive has been requested to reply directly to you in relation to the above parliamentary question, which you submitted to the Minister for Health for response. I have consulted with the National Heart Programme (NHP) on your question and have been informed that the following outlines the position.

Transthyretin (TTR) Amyloid is an increasingly recognised and serious, potentially fatal condition where abnormal protein deposits accumulate in organs and tissues, most commonly the heart (ATTR-cardiomyopathy, ATTR-CM) and the nervous system. It can be inherited and run in families but more commonly it occurs in older patients >65 years without evidence of familial inheritance (wild-type ATTR-CM). It is estimated to be present in 1-3% of the European population over the age of 70 years.

The condition is being increasingly recognised due to the availability of non-invasive imaging techniques to diagnose it and the emergence of several disease modifying therapies which have completely transformed the course of the disease. The key priority is to diagnose patients with the condition early enough so that they can be assessed for and prescribed these therapies in a timely fashion, to improve outcomes.

Recently, disease modifying therapies for the hereditary and principally neurological form of the disease have now been proven to have significant benefit also for the wild type and cardiomyopathy form of the disease. This is likely to have significant implications for numbers of patients eligible for these treatments.

Individuals with TTR amyloidosis affecting their heart are best managed by cardiologists with an interest and expertise in managing this condition, with access to diagnostics tests including imaging and genetics services, working together with subspecialist neurologist/neurophysiology consultants particularly in the case of the hereditary disease. Diagnosis can be delayed in the absence of this subspecialty awareness and referral. This is principally because the symptoms may mimic other more common diseases and affected patients may attend many different doctors before the diagnosis of amyloidosis is made.

Disease-modifying treatments for ATTR-CM which improve quality of life and prognosis despite their high costs are approved for reimbursement in Ireland under a Managed Access Protocol, and ideally, are prescribed and monitored by specialised clinicians. The Mater Hospital has recently been developing and growing its well-established focus on diagnosing, treating and monitoring ATTR-CM, aiming to address the specific needs of patients with amyloidosis.

The HSE Amyloidosis Model of Care was approved in 2022. Since then, there has been progress with the approval of pharmacological treatments included in this model of care, where relevant, through the HSE's medicines management approvals process.

When a model of care is approved it is then progressed to implementation operationally. There were a number of areas identified in the model of care that required funding in order to progress to implementation. Funding was sought to support phased implementation of this model through the HSE's national service planning process with the Department of Health in 2023. Unfortunately, this new service development submission did not secure funding at that time.

This submission would have been part of a number of submissions for funding for new service developments and final allocations for such developments would have been outlined in the Letter of Determination from the Minster of Health to the HSE.

However funding has been allocated to rare diseases in National Service Developments (NSDs) for 2025 and it is planned to consider how some of this funding could support Amyloidosis.

I trust this information is of assistance to you but should you have any further queries please do not hesitate to contact me.

Yours sincerely

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Anne Horgan
General Manager