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26 September 2025

Deputy Ciarán Ahern, TD
Dáil Éireann
Leinster House
Kildare Street
Dublin 2

**RE: PQ 49659/25**

**To ask the Minister for Health if her attention has been brought to Okur-Chung Syndrome, an ultra-rare genetic disorder caused by mutations in the CSNK2A1 gene which affects neurodevelopment, and of which there are seven cases in Ireland; if she is aware of any research into the disorder; if not, whether she will consider having such research commissioned; and if she will make a statement on the matter.**

Dear Deputy Ahern,

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 Rare Diseases Office on your question and have been informed that the following outlines the position.

Okur-Chung syndrome is genetic condition caused by pathogenic (disease-causing) variants in the CSNKA1 gene which has an important role in neurodevelopment. It is an ultra-rare condition an estimated prevalence of less than 1 in a million. People with Okur-Chung syndrome commonly have developmental delay, intellectual disability, ataxia, seizures, musculoskeletal problems and issues with speech and language, behaviour, feeding and sleep.

The centres of expertise in Ireland for neurodevelopmental conditions such as Okur-Chung syndrome are the Child Development and Neurodisability services in Children's Health Ireland at Tallaght, Crumlin and Temple St. For adults Tallaght University Hospital and Beaumont Hospital provide subspecialty clinics including movement disorders, cognitive, neuromuscular and epilepsy which may be relevant to patients with Okur-Chung syndrome. Services are also provided in other neurology centres.

National research initiatives for ultra-rare conditions can be challenging due to the small numbers of people with each condition. Therefore, research studies often involve cross-border or international collaboration with families and clinicians.

Ireland has been a key participant in the Deciphering Developmental Delay (DDD) research study which aims to identify the underlying genetic cause of developmental delay in children. A number of Irish patients were diagnosed with Okur-Chung syndrome through participation in DDD which has enabled better understanding of the condition.

European Reference Networks (ERNs) are networks of healthcare professionals across the EU to enable specialists to share knowledge on the diagnosis and management of rare complex conditions and promote opportunities for research. Ireland is a full member of ERN ITHACA (rare intellectual and neurodevelopmental disorders), ERN-RND (rare neurological disorders) and ERN-NMD (rare neuromuscular disorders).

The international CSNK2A1 foundation, established in 2018, aims to connect families with each other and promotes research.

Simons Searchlight is an international research group with a focus on rare neurodevelopmental disorders. It gathers information from families to improve knowledge of each condition. Okur-Chung Syndrome/ CSNK2A1 gene is listed as a condition they study. Families can register directly with Simons Searchlight to participate: https://www.simonssearchlight.org/research/what-we-study/csnk2a1/

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

**Anne Horgan**
**General Manager**