

Centralised Efforts Transforming Global Care

The Ten-Year Impact of a Patient Advocacy Group – Medical Expert Partnership on Recognising and Treating a Rare and Complex Epileptic Brain Malformation Syndrome

Introduction

Hypothalamic Hamartoma with Epilepsy (HH) is an under-recognized, rare and congenital brain malformation syndrome. Gelastic seizures, the hallmark seizure-type, are often missed.

As recently as 2010, recognition and treatment of this syndrome was limited to pockets of expertise in diverse global locations. Even within these centres, patient follow-up and research lacked global oversight.

Our patient advocacy group, Hope for Hypothalamic Hamartomas, has been transformative in uniting and streamlining the work of these diverse clinical and scientific institutions, initiating and enabling co-operation for mutual benefit.

Methodology

Hope for HH transforms patient outcomes through its diverse, expert [Medical Advisory Board](#). We host expert International Symposia focused on progressing critical research, and fund and support [cross-border research partnerships](#). Our aim is to increase understanding of this rare syndrome to improve patient management and treatment, and provide avenues to develop new therapies.

“ From the point of view of our work in Melbourne, I think particularly for a rare disease like HH (incidence ~ 1 in 200,000), that the most important thing Hope for HH has done is maximise the number of patients and families being involved in research. As you know, research - and particularly genetics - is often a numbers game. And you and the other families have certainly maximised our numbers! ”

Michael Hildebrand, geneticist

“ Our multidisciplinary group dedicated to the management of severe drug resistant epilepsy started to manage HH patients in the early 90's and we introduced radiosurgery to our armoury in the late 90's. Due to the rarity of the disease, the lack of knowledge of the majority of practitioners was a significant obstacle to early diagnosis, to referral for surgery and to proper follow up of patients coming from distant countries. Although I had joined the inaugural Medical Advisory Board of Hope for HH in 2011, it was the Marseille Symposium in 2013 that really brought home to me the importance of collaboration with patient advocates and the impact that this can have on the sharing and development of expertise. Since then, through a collaborative approach Hope for HH has capitalised on the impetus created at the Marseille symposium. I have no doubt that very many patients condemned by incorrect diagnosis, and then sinking into an irreversible epileptic encephalopathy with severe cognitive and psychiatric comorbidities, have been given, thanks to Hope for HH, the chance to have again a future! I have no doubt that this collaboration has dramatically improved our capacity to progress in the fight against this complex disease and has directly transformed the future of hundreds of these patients and their families. ”

Jean Régis, neurosurgeon

Key outcomes

1 Connecting patients and families

By establishing the advocacy group and encouraging members to network with each other, participate in research, and drive research funding initiatives. This has maximised participants for global research programs, vital for a rare disease like HH.

2 Driving Understanding

International Symposia, held every three years, provide specialists with an invaluable platform for discussion between peers across institutions and disciplines, and also enable those specialists to receive guidance from the patients and families as to how care might be improved. These symposia have been described by [expert participants as 'priceless'](#) and ['critically important'](#) resources to further understanding, to enable consideration of the global syndrome with all its comorbidities, and to drive research.

3 Promoting Treatment

Surgical techniques that were nascent in 2010 and limited to a handful of countries have been, through this patient/professional partnership, developed, refined and disseminated internationally, giving hope to patients wherever they live. Urgent diagnosis and surgical intervention are vital for HH patients since the seizures are often subtle but refractory to medication; delays can lead to intellectual disability and behavioural disorders that become increasingly unmanageable. Evidence that patient collaboration is leading to earlier diagnosis and effective surgery may derive from the younger average age of surgical patients now reported by the treating centres. Research is continuing to determine the comparative effectiveness of surgical techniques.

4 Developing Guidelines

The partnership has led to development of the first guidelines and recommendations for clinical management of patients - the ['HH Clinical Pathway'](#), - now accepted as best practice at most specialist epilepsy centres. Expert committees chaired by Medical Advisory Board members are currently working on the creation of clinical guidelines for diagnosis and treatment of HH within four categories: endocrinology, psychology/psychiatry, neurology and imaging.

5 Supporting Research

Cross-border research initiatives, such as [a genetic study](#) run by The University of Melbourne, with critical input from centres in the US, Europe and Japan, is promoting greater understanding of aetiology, moving us closer to new therapies for patients refractory to surgical treatment.

6 Targeting Co-Morbidities

Through patient advocate input, focus has widened beyond surgical outcomes. HH is now regarded as a complex syndrome requiring a multidisciplinary approach and cross-specialty cooperation. For example, counselling and psychological support are increasingly being recognized as beneficial for patients and their families. Improved quality of life sits alongside seizure control as a primary treatment goal.

7 Sharing Knowledge

The partnership has developed comprehensive website and social media platforms through which the patient community can access support and information.

Conclusion: Through mutually beneficial partnership, patient advocates and medical professionals can promote international cross-specialty relationships which enrich medical understanding and transform patient care.