Diagnostic, neuropsychiatric and therapeutic considerations in epilepsy and intellectual disability

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Valorisation

Relevance of findings

It is not exactly clear how many people with intellectual disability (ID) live in the Netherlands. According to calculations by the Sociaal Cultureel Planbureau (SCP), in 2013 there were around 142,000 people with ID. Additionally, there are approximately 1.4 million people with a borderline ID (IQ between 70 and 85). The expenditure on the care for people with ID amounted to around 7.3 billion euros in 2015, 8.6% of the total healthcare costs in the Netherlands.¹

People with ID have much higher rates of epilepsy than the general population (22.2% vs 1%)²,³, their epilepsy often is more difficult to diagnose and treat, it is more likely that treatment requires polypharmacy and patients may find it harder to communicate their wishes and needs.⁴

People with ID and epilepsy are part of a vulnerable population in society, whom deserves the best healthcare. But care and treatment pathways for this complex subpopulation are poorly defined and there are no strict specific national or international guidelines available yet.

Epilepsy and ID have been linked to a variety of behavioral, affective and psychiatric comorbidities⁵-⁷, but we also know that epilepsy a condition which also often falls into the mind-body gap in terms of the way clinicians or services respond⁸. This is one of the motivations to carry out this PhD in a “twin”-form, that stresses the relevance of a multidisciplinary and holistic approach. This thesis covers the medical perspectives, the other thesis is performed by a behavioral scientist.

These theses are an effort to make a pragmatic, yet evidence-based, bridge over the yawning gap of available literature and challenges faced in daily clinical practice.

In the first part of this thesis we showed that unraveling the etiological diagnosis could be beneficial to both adult as well as pediatric patients, and should be considered at any age and any patient. By using whole exome sequencing (WES) we were able to find an explanation in more than 25% of patients, and in over 50% of these patients this diagnosis might even be relevant for treatment strategy in the future.

In a population in whom the combination of epilepsy and neuropsychiatric problems are common, cause and effect are often difficult to disentangle, but our thesis reveals that the role of epilepsy characteristics seems to be modest compared to ID-characteristics. This shows that there should always be attention for other factors besides epilepsy-related problems.
Before any medication introduction, stop or change it would be helpful to document baseline behaviors in a more standardized way, in order to monitor changes during follow-up. Publishing our evaluation of such an approach to psychiatric symptoms would therefore be beneficial for both the patient and the clinician.

The second part of this thesis consists of relevant clinical findings concerning the therapeutic options in this complex population. Over the past few years, various AEDs with new modes of action were introduced, but patients with ID and epilepsy are often excluded from the initial registration studies, as they are complicated for both practical and medical ethical reasons. We show that, although challenging, newer AEDs should not be avoided when facing problems in the current therapy, but the refractory nature of epilepsy should be part of counseling families so they are informed on realistic grounds.

Our last chapter shows that even when theoretically an AED should be avoided based on a specific etiology or diagnosis, withdrawal should be very closely monitored by an experienced neurologist or clinician who is aware of both the risks and benefits for this specific patient.

**Target groups**

The principal purpose of this thesis is to help colleagues working with people with ID and epilepsy to increase their knowledge with regard to evidence based assessments, investigations and ultimately also treatment choices in this complex population. We hope to encourage them to take an active role in the attempt to enhance the evidence-base of considerations and treatment choices.

With increasing the knowledge and awareness of the healthcare professional, the indirect target groups of course are the patients and their relatives.

**Activities for innovation and implementation of knowledge**

All studies in this thesis are published or submitted as scientific manuscripts in peer-reviewed international journals. Several findings have also been published in national peer-reviewed journals.

We participated in national and international congresses and symposia, also reaching out to non-academic healthcare professionals.

Additionally we included a large part of the outcomes in training programs and seminars for both doctors and nursing staff.
Different findings of this thesis will be implemented and considered for the national epilepsy guideline by the “Nederlandse Vereniging voor Neurologie” (NVN).

Furthermore, this thesis resulted in a new national collaboration with the CCE (“Centrum voor Consultatie en Expertise”), bringing experts in the field of behavioral problems and epilepsy together in a multidisciplinary team which can be consulted throughout the whole of Netherlands.

Our international collaboration with the UK will be expanded to other countries, making more and more use of existing datasets. This enhances the opportunity to compare the different experiences from international centers in order to establish an international guideline that is, in line with the white paper⁸ focused on this specific population. International studies with a prospective design are part of the ‘strategic nota’ for 2020–2024 of our research-team. Since 2019 we are also part of the ILAE taskforce for people with ID and epilepsy.

Future plans include a website and/or app for patients and caregivers, which can be filled with results and alerts coming from different studies in the future, with which the pathway of considerations can be expanded. It is important to keep in mind that this pathway should be interpreted as a continuum rather than a definitive categorical structure, and the results of this thesis are just the primary pieces of the puzzle.
References