Conclusions: These new findings suggest that the permanent neuronal damage was not found in the patients with breath holding spells. Additionally, our results were demonstrated that increased glutamate levels certainly may play a role in the risk for developing neuronal hyperexcitability in these children.

p0343 CLINICAL PARAMETERS IN CHILDREN WITH CEREBRAL PALSY AND EPILEPSY

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Purpose: Cerebral palsy (CP) is characterised by chronic nonprogressive neurological disorders of motor function, posture and movement. The purpose of this study was to determine the clinical parameters in children with CP and epilepsy.

Method: This retrospective study included 31 children with CP who were treated at the Clinic for Neurology and Psychiatry for Children and Youth. In all children were analysed the clinical form of CP, type of epileptic seizures, Apgar score, the presence of neonatal epileptic seizures, ultrasound (US) of the brain and electroencephalographic (EEG) results in the first year of life and positive family history for epilepsy.

Results: Epilepsy developed in 10 children with CP, and 7 children had partial seizures with or without secondary generalisation, and 3 had infantile spasms. In the group with epilepsy, 9 children had hemiparetic and quadriparetic form of the disease, and 1 child had diplegic form. Neonatal seizures and abnormal EEG showed statistically significant association with occurrence of epilepsy in children with CP (x^2 = 5.9, DF=1, p < 0.05; x^2 = 5, DF=1, p < 0.05), while US of the brain (x^2 = 2.8, DF=1, p > 0.05) and low Apgar score (t = 0.183, p > 0.05) and positive family history for epilepsy (x^2 = 3.1, DF=1, p > 0.05) did not show such significant association.

Conclusion: Neonatal seizures and early EEG are good predictors for later occurrence of epilepsy in children with CP, while US of the brain, Apgar score and positive family history for epilepsy are not such a good predictor. The most common type of CP associated with epilepsy were hemiparetic and quadriparetic type.

p0344 PAEDIATRIC OFF-LABEL USE OF ANTI-EPILEPTIC TREATMENT IN INTRACTABLE CHILDHOOD EPILEPSY: A SURVEY IN A LARGE COHORT OF PATIENTS WITH DRAVET SYNDROME

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Purpose: The aim of this study is to explore the extent of off-label use of antiepileptic drugs (AEDs) in a cohort of patients with Dravet Syndrome (DS), a pharmacoresistant epilepsy.

Method: We proposed auto-administered semi-close on-line survey to families of pediatric patients with DS in December 2013 on the website of DS alliance, France. Questions addressed different aspects of patients' medications in particular the off-label including all aspects of drug prescription not included in the SPC: therapeutic indication, posology, pharmacological form and route of administration.

Results: 89 families answered the questionnaire. Patients were aged from 0.84 years to 17.60 years (mean 8.06). The most used AEDs were the following:

- clobazam (92%, CLB)
- valproate (90%, VPA)
- stiripentol (81%, STP)
- topiramate (46%, TPM)

We reported the major category of off-label use for these four drugs:

- 28% of patients use CLB out of the age-subsets,
- All patients with TPM are not in therapeutic indication for DS,
- 35% takes STP in a mixture non indicated with this treatment,
- The usual posology is not appropriate for TPM, VPA, CLB and STP respectively in 93%, 66%, 52% and 21% of patients,
- The dosage for age is not appropriate for CLB, STP, VPA and TPM respectively in 57%, 26%, 11% and 10% of patients,
- Care givers should manipulate CLB, STP, VPA and TPM in respectively 63%, 31%, 13% and 12% of patients.

Conclusion: This study raises the difficulties in families with children with DS using often off-label AEDs polytherapy. These results emphasize the need for further development and studies of paediatric formula of drugs currently used in Dravet Syndrome.

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National Registry of Dravet’s Syndrome and other Syndromes correlated with genes SCN1A and PCDH19

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**01** INTRODUCTION

Patient Registries have been recognized by the European Community as one of the principal tools in the prevention of lasting interventions in the frame defined by the Tissue and cell bank directive. To improve the understanding of the disease through the systematic and continual registration of data for basic and epidemiological researches, with the aim of developing care therapeutic solutions, the National Registry of Dravet Syndrome (NDR) was established in 2008. The DR Lee Foundation (http://drell.it) has promoted the development of the National Registry of Dravet Syndrome and other Syndromes related to mutations on SCN1A and PCDH19 genes.

**02** OBJECTIVES

Dravet Syndrome (DS), also known as severe myoclonic epilepsy of infancy, is a rare form of epilepsy associated with neurological developmental disorders. The incidence of the disease has been estimated between 1/20,000 and 1/25,000, with a greater frequency in males than in females. The incidence of the syndrome is unknown in the general population, but it is considered extremely rare. The purpose of the Registry is to improve the scientific community, contribute to the identification of patients, in order to collect and make available a uniform source of data concerning DS and related conditions. The Registry provides an important source of information on clinical and laboratory findings, treatment, and the interaction of all research and sanitary structures present in the country, in support of more adequate strategies for diagnostic, therapy and care.

**03** METHODS

The writing group, after having identified the main areas of the registry, elaborated an accurate protocol, including an approach, a description of data, a template, a case definitions, a case history, a registry, a registry, a registry, a registry, a registry, a registry, a registry, and other sources. The research work was performed by the Dravet Italia Onlus (http://drell.it) and other sources. The information regarding the registry has been validated in order to collect the information according to the American standards and appropriately. For the definitions of drug therapy, name the name of an international form has been used.

**04** RESULTS

Preliminary data. Until now, we have collected information for 258 patients of which 87 affected by Dravet syndrome (DS) (48 females, 39 males), and 50 (36 females, 24 males) affected by Other Syndromes (OS) with SCN1A gene mutation, 61 (27 females, 34 males) affected by PCDH19 gene mutation but without seizures. 18 PCDH19 gene mutations (18 females).

**05** EXPECTED RESULTS

Analysis of clinical, genetic, and epidemiological data regarding these syndromes. Evaluation of therapeutic treatments efficiency. Evaluation of social, psychological, and behavioral outcomes.

**06** CONCLUSIONS

The Registry is a powerful instrument to support the improvement of the understanding of DS-related syndromes, in order to improve the standardization of the therapy and promote a network research. The Registry could represent an important tool for the systematic collection of data from all patients, in order to promote the knowledge of the disease, and its nosological and genetic aspects will be made available to interested parties, for promoting and sustaining scientific research, with the aim of discovering innovative therapeutics options for the management of DS and related syndromes. Drawing from these preliminary results, we would like to expand the use of the registry in Europe.

**07** BIBLIOGRAPHY