Grant agreement no. 769051

EpiCARE – a network for rare and complex epilepsies

HP-ERN-2016 European Reference Networks / Framework Partnership Agreement

D15.1: Proposal for EpiCARE clinical database

Work Package: Research IV (WP15)

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1. Version log

Version	Date	Released by	Nature of Change
01	8th January 2018	R. Guerrini (Meyer Hospital)	First version
02	31st January 2018	R. Guerrini (Meyer Hospital)	Deliverable report was modified in order to make it clear what information is collected in the dataset

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2. Definition and acronyms

Acronyms	Definitions
EpiCARE	European Reference Network for rare and complex epilepsies
RedCap	Secure web application for building and managing online surveys and databases
TC	Teleconferences
Epi25K	Large-Scale Whole Genome Sequencing in Epilepsy consortium

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3. Introduction

Research based on clinical practice is an important component of EpiCARE. A database, as reported by the deliverable D 15.1, is the starting point for (clinical) research projects promoted by EpiCARE members, as it may indicate how many patients with a specific rare and complex epilepsy can be enrolled and can also facilitate coordinated efforts for applications to Horizon 2020, E-rare calls, clinical trials and publications on even extremely rare disorders.

The deliverable D 15.1 refers to objective 5 of EpiCARE proposal: “To enhance opportunities for registries, and collaborative research for the benefit of individuals with rare and complex epilepsies across Europe. EpiCARE utilizing the database/registries may allow natural history studies of individual phenotypes (to include epidemiologic, phenotypic, genotypic, response to medication, natural history, comorbidity, long term outcome, existing treatments, mortality) and determine further key areas of research priority to be addressed, as illustrated with the Dravet syndrome experience, for example (http://dravet.eu/).

The specific objective of deliverable D 15.1 is: “to establish a European clinical database (REDCap) for complex and rare epilepsies”. A database is an assortment of data that is organized to be easily accessed, managed and updated. Such a research prone, clinical database will subserve clinical trials, genetic and epidemiologic research.

4. Activities carried out and results

The structure of the clinical database reported in the deliverable D 15.1, has been set up through discussions in the context of Steering Committee monthly TC and dedicated face-to face meetings. Prof Lieven Lagae, WP I leader, has significantly contributed to the development of the database.

As first step, differences between registries and databases have been pointed out. Secondly, the Epi25K database (http://www.epgp.org/) has been chosen as a model to build the EpiCARE database, and has been adapted to more general clinical purposes and simplified to make it particularly suitable to transfer already stored data and quickly collect new data even with limited resources available. In order to feed the data in reasonably quick way, the database has been simplified and a modular structure has been used.

An agreement has been reached concerning the minimum dataset (i.e. demographics, etiology and syndromic classification of epilepsy, comorbidities and treatment) during a dedicated Steering committee meeting (3rd December 2017). This scheme has been

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circulated to the Steering committee members for further amendments in December 2017.

Finally, a clinical database fulfilling D 15.1 requirements has been set up (see D15.1 ANNEX 1). As shown in the D15.1 ANNEX 1, this database contains core (clinical) data (limited number of relevant data fields: demographics, age at epilepsy diagnosis, etiology of epilepsy, type of epilepsy syndrome, presence of associated intellectual disability, current medical and dietary treatment, previous surgical treatment) with drop down menus for specific items (i.e different types of epilepsy syndromes and current medical treatment) and does not need to be updated very frequently. It is modular, using the REDCap platform, e.g. the core data set (the core module) is common to all patients, but each center or patient group can add (locally) other data fields for its own use.

The major achievement of this deliverable is the proposal of a research prone clinical database which will hopefully include all available patients through participation of HCPs and patient groups included in EpiCARE.

Pending issues are as follows:

1) anonymization process (the database should be provided with anonymized codes so that duplication of data by different centers would be impossible),
2) accessibility (the database should be made accessible only after duly signed informed consent forms have been approved and adopted by all centres)
3) ethical approval by local boards

5 Conclusions

A proposal for a research prone clinical database has been set up through consensus in the context of the EpiCARE Steering Committee. This database represents the first step to coordinate efforts for applications to European research calls for the benefit of individuals with rare disorders, which is one of the main objectives of EpiCARE.

6 Bibliography / References

Lanthaler B, Wieser S, Deutschmann A, Schossig A, Fauth C, Zschocke J, Witsch-Baumgartner M. Genotype-based databases for variants causing rare diseases. Gene. 2014;550(1):136-40.

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ANNEX 1.

EPICARE clinical database

CORE MODULE

informed consent given	yes/no		European new consent form : 3 parts
Demographics	unique patient identifier		
	date of birth		
	sex male female		
	dead/alive		specify date and cause of death (SUDEP?)
date database entry			
date last update entry			
current age	years/months/days		automatic calculation: current date - birth date
age epilepsy diagnosis	years/months/days		for children< 1year and neonates months and days are possible
etiology of the epilepsy	structural	MRI available?	specify (possibly with drop down menu)
	genetic	DNA available?	specify
	metabolic		specify
	immune		specify
	infectious		specify
	unknown		
epilepsy syndrome	not known		
	known		drop down list, see epi25K
encephalopathy	yes/no		
intellectual disability	yes/no		
treatment	AED		specify generic names (drop down list)
	ketogenic diet		start date / stop date if stopped
	VNS		specify implantation date
	surgery		specify date and type of surgery

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