

The European Prader-Willi Syndrome Clinical and Basic Science Research Project

Prader-Willi Syndrome: a model linking gene expression, obesity and mental health

www.pwseu.eu

Background to this research project

An integrated multidisciplinary investigation of Prader-Willi Syndrome (PWS) includes:

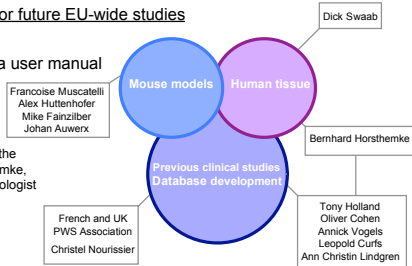
- Molecular biological studies of human tissue and knockout mouse models of PWS**
 - Establishment of genetic mouse models
 - Investigation of neurobiological mechanisms and signalling pathways
 - Study of human hypothalamic tissue
- Development of a clinical research database for future EU-wide studies**
 - Internet-based PWS research database
 - Testing of the database and development of a user manual

Clinical partners:

Olivier Cohen HC Forum (France); Leopold Curfs, psychologist (the Netherlands); Tony Holland, psychiatrist (UK); Bernhard Horsthemke, geneticist (Germany); Ann-Christin Lindgren, paediatric endocrinologist (Sweden); Annick Vogels, child psychiatrist (Belgium)

Other participants (database development):

Christel Nourissier, PWSA (France); Michael Boasis, HC-Forum (France); Oenone Dudley (France); Joyce Whittington (UK)



Clinical research project

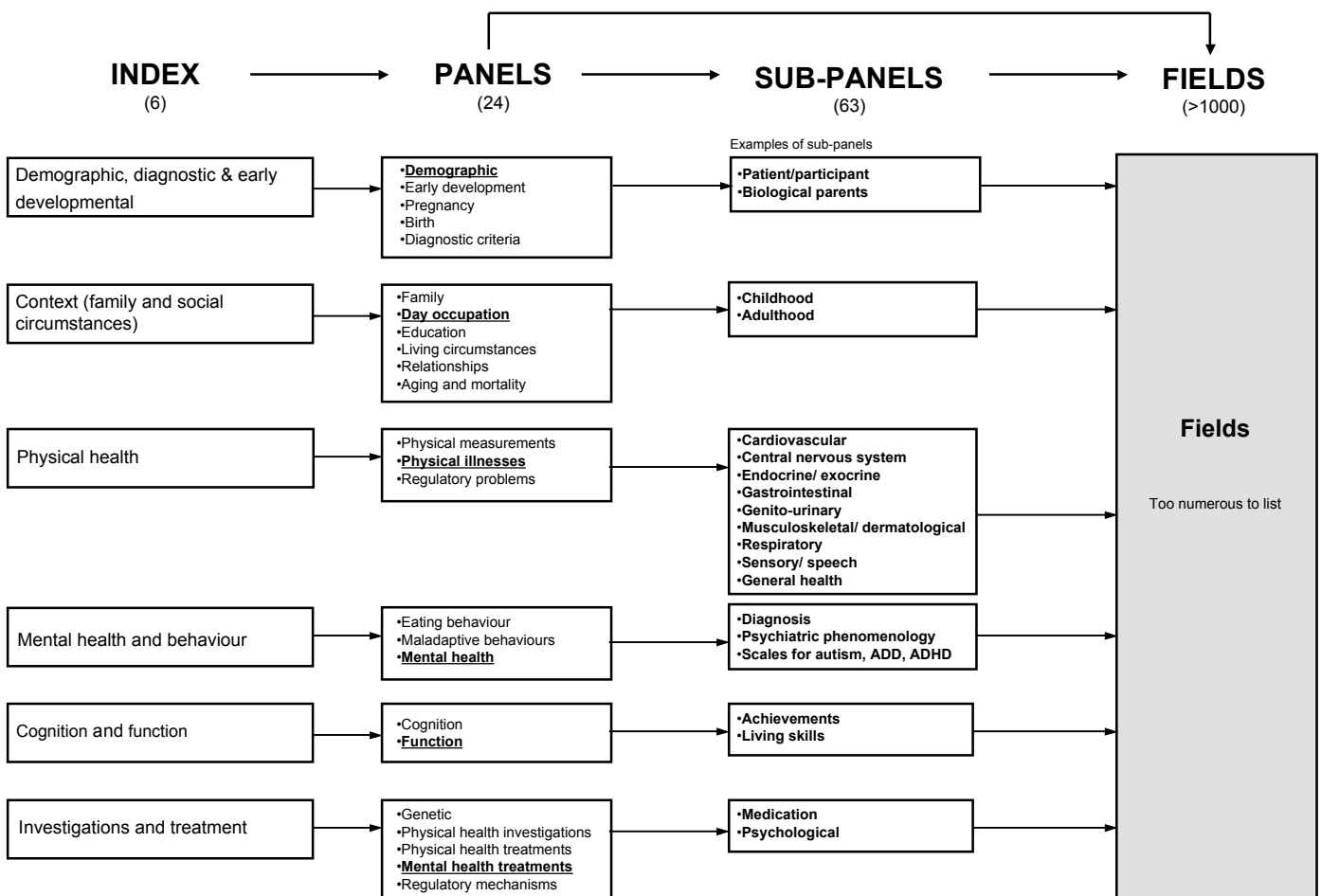
Why have multi-centred international clinical studies?

- To investigate influences on the physical, cognitive and emotional developmental trajectories of people with PWS across their lifespan
- Where analyses are problematic due to small sample sizes in any one country when many variables need to be considered (i.e. age, genetic sub-type etc.)

Benefits of an EU clinical research database

- To enable the reliable and lawful collection, management and analysis of data across many countries thereby having sufficient numbers of people with PWS to answer specific questions
- To enable comparisons between countries allowing the influence of different educational, health and social care policies on the lives of people with PWS to be investigated
- Through the use of the database to undertake research to establish a critical mass of inter-disciplinary clinical expertise and to enable the development of best practice across the EU

Structure and size of the database



A schematic of the database showing selected panels (in bold) and their sub-panels

Timetable

- Completion of the database: *September 2007*
- Trial of the database: *September 2007 - March 2008*
- Development of the database manual: *May 2007 - November 2008*
- Establishment of the database administrative structures: *September 2007*

Future developments

- Establishment of partnerships between national PWS Associations and research groups in each country
- Identification of countries with established partnerships wishing to undertake collaborative studies
- Identification of research that requires national collaborations to be successful
- Applications for funding

