

Sixth Framework Programme

Life Sciences, Genomics and Biotechnology for Health
Specific Targeted Research Programme

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

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www.pwseu.eu

Outline

- Background
- Reasons for the database
- Basic structure
- Demonstration of the database
- Rules for database use
- Database Management Group
- Long term support for the database
- Collaborative studies with existing data

The need for multi-centred, international studies

- Influences on the physical, cognitive, and emotional development trajectories of people with PWS across their lifespan
- Population prevalence 1: 54,000
- Sub-divisions
 - Genetic sub-type
 - Other genetic factors
 - Gender
 - Weight control
 - Early intervention
 - Growth and/or sex hormone replacement etc

Why develop an EU database?

- To enable the reliable and lawful collection, management and analysis of data obtained using agreed methods across many countries;
- Comparisons between countries to enable the influence of different educational, health and social care policies on the lives of people with PWS to be investigated;
- Establishes a critical mass of inter-disciplinary clinical expertise to focus on a rare disorder and the development of best practice across the EU.

Agreed objectives

- To facilitate multi-site, clinical research on PWS
- To standardised assessment procedures so data can be combined between groups and over time
- To reduce the need for different groups to collect the same data
- Accumulated data to allow the testing of novel hypotheses and to enable longitudinal studies
- Cross-cultural and cross-national studies

Support for multi-centred studies

EU Project 6th Framework

Aims

To develop a database that will enable multi-national clinical studies of PWS

Database must be:

- Compatible with EU and national laws
- Accessible from multiple sites
- Support the storage, management, analysis and presentation of cross-sectional and longitudinal data

General Issues

- Primarily clinical or research use?
- Specific PWS focus?
- Reliability of data collection (issues of language)?
- How extensive to be?
- Management of database at national and supra-national levels

Structure & content of the database

Index Panels Sub-panels Fields

Index

- Demographic, Diagnostic, Developmental
- Context
- Physical health
- Mental health and behaviour
- Cognitive and functional abilities
- Investigations and treatments

Content of the database

- **INDEX**
 - Physical Health
- **PANELS**
 - Physical measurements (fields include Ht, Wt, BMI)
 - Physical illnesses (see sub-panels below)
 - Regulatory problems (fields include sleep apnoea etc)
- **SUB-PANELS**
 - Cardio-vascular
 - Central Nervous System
 - Endocrine/exocrine
 - Gastrointestinal
 - Genito-urinary
 - Musculo-skeletal/dermatological
 - Respiratory
 - Sensory/speech
 - General health screening
- **FIELDS**

Demonstration of the database

Use of the database

Database Manual

- Integrity and confidentiality of personal data
 - Consent procedures
 - Anonymous data
 - Long term use of data
- Quality of data stored on the database
 - Quality of data collection
 - Assessments used
- Combining of data
 - Collaborations
 - Anonymous data
 - Publications

Rules of use

Database Manual

- Users of the database must have adequate consent procedures
 - Specific use
 - Combined use
 - Long-term use
- Database to enable a research group to store data exclusively for their use – identity of participants confidential to them
- Agreed protocols for the sharing of data in an anonymous manner
 - Collaborative hypothesis driven research
 - Exploratory novel investigations

Database Management Group

- 'Development' group to 'Management' group
- Integrity of the database
- Applications for use of the database
- Oversee publications arising from the database
- Strategy for long-term use
- Removal of data
- Resolve disputes

Long-term support

- At national level
 - Research groups
 - National PWS Associations
- At EU level
 - Rare diseases infra-structure
- At international level
 - ?IPWSO

Possible UK Model

Seek grant funding for:

- Establish database support at UK PWSA
- National database management group chaired by representative from UK PWSA
- Presentation and demonstration of database at PWSA meetings
- Contact of past and present recruits with PWS
 - New consent procedures
 - Up-date of recent clinical information
- Recruit new participants
 - Core clinical information
- Establish long-term recruitment strategy

Testing the database through collaborative studies

- Hormone replacement
- Mental health
- Ageing

Unresolved questions?

- Who owns the database?
 - Software
 - Content
- Long term support and costs?
- Long term management?
- Commercial involvement?