

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

Aims of project

An integrated multidisciplinary investigation of Prader-Willi syndrome (PWS)

- 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
- Establish the basis for future EU wide clinical studies by developing a clinical research database
 - Internet-based PWS research database
 - Testing of database and development of user manual

Workpackages

- WP1 Management
- WP2 Clinical study
- WP3 Genetic Study
- WP4 Basic science
- WP5 Integration
- WP6 Dissemination

WP 1

Management

Within first year

- Management structure
- Ethical approval
- Website

By 42 months

- Final Plan for dissemination
- Raising public awareness
- Final reports

WP2

Clinical Project

By 18 months

- Content of database
- Rules for use
- Database

By 42 months

- Trial of database in three countries
- Establish the future of the database

WP 3

Genetics

By 18 months

- Establish diagnostic criteria for database

By 42 months

- Frequency of snoRNA gene deletions in 'PWS-like' patients
- Clinical description of patients with IC defects
- Frequency of IC deletions

WP4

Basic science

By 42 months

- Establish a minimum of 3 mouse models
- Phenotypic and physiological assessment of all 3 models
- Identification of in-vivo cellular defects
- Identification of relevant pathophysiological and signalling pathways for candidate genes
- Investigation of systems in the hypothalamus – at least one new feeding pathway
- Validation of identified candidate genes from the mouse models

WP5

Integration

By 3 months

- Timetable for forum for exchange of ideas

By 42 months

- Identification of similarities and dissimilarities between PWS and mouse models
- Identification of potentially new PWS genes
- Develop new models of understanding of genotype/phenotype relationships

WP6

Analysis and dissemination

By 42 months:

- Publication of scientific papers
- Use of database in different countries
- Publications in practice-based journals and through National Associations
- Research policy for rare disorders
- Development of a website for the lay public and PWS Associations

Plan for dissemination

- List of papers from different groups to be added to website;
- Lay summary of basic science and clinical project to be prepared and added to website and sent to relevant PWS Associations for inclusion in newsletters;
- Relevance for rare disorders research
- Final report

Future research

- Basic science
- Clinical