

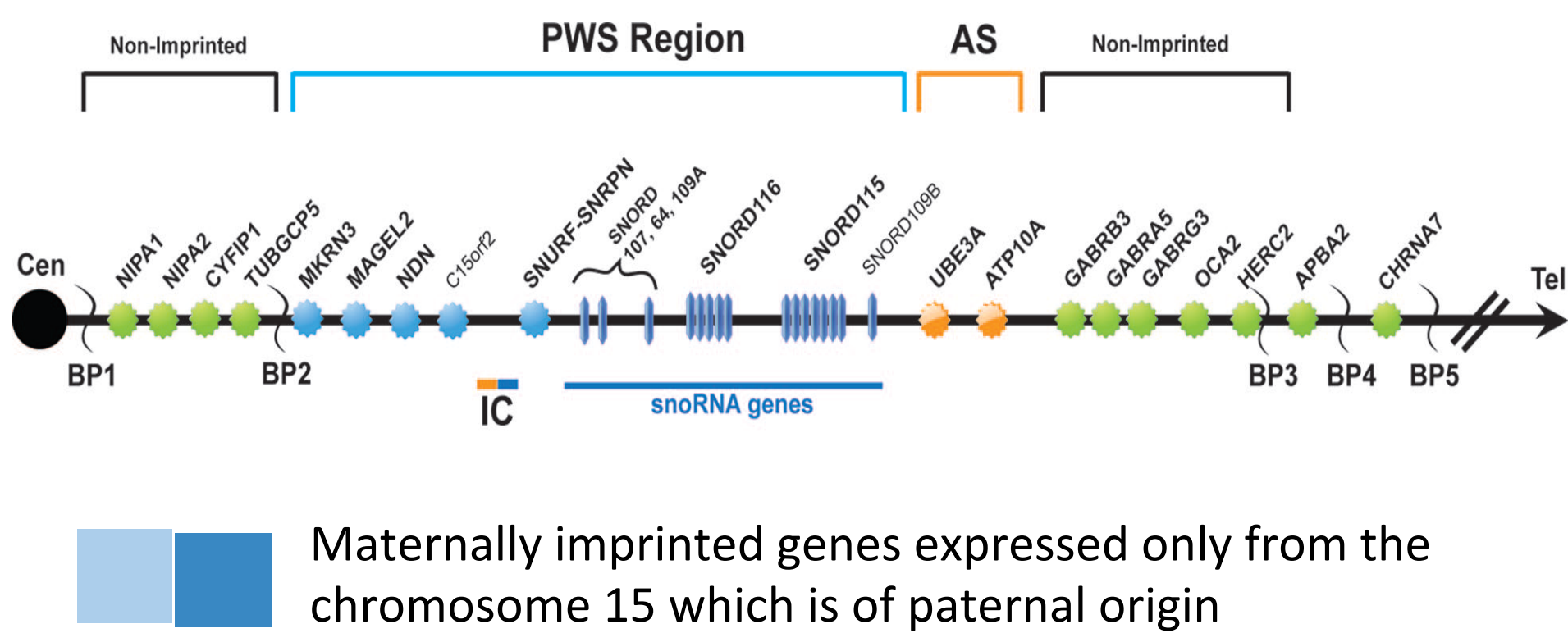
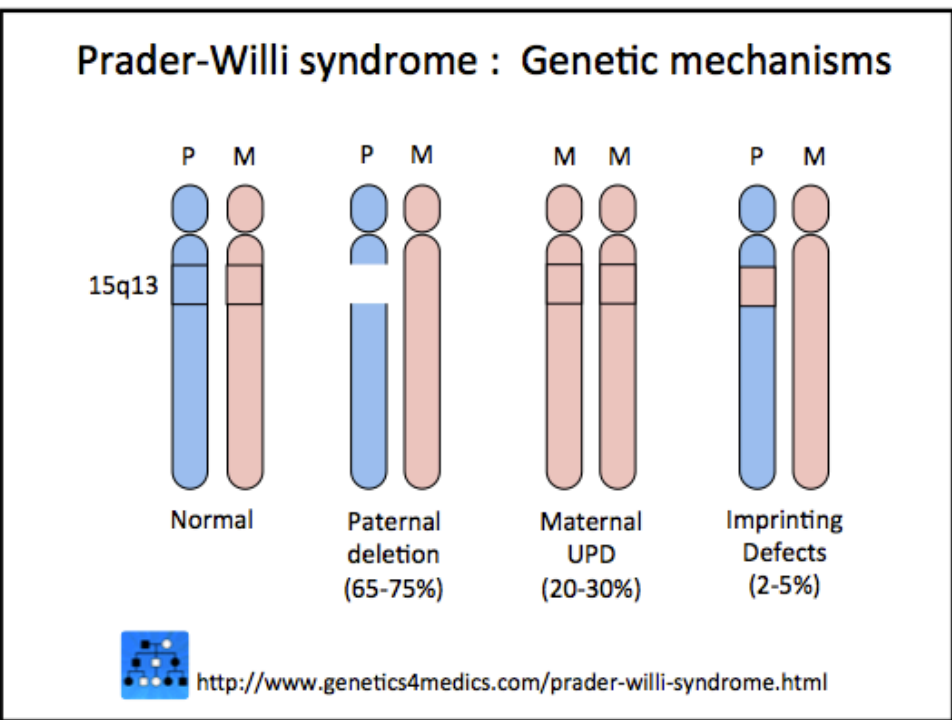
Addressing the Ethical and Therapeutic Challenges of a Rare Disability: Hyperphagia & Other Behaviours in People with Prader-Willi Syndrome (PWS)

Tony Holland*, President IPWSO, Emeritus Professor, Cambridge University, UK; Maria Libura, IPWSO Trustee, President Polish PWSA; James O'Brien, IPWSO Trustee, President Australian PWSA; Elizabeth Fistein, Specialty Director in Ethics and Law, Clinical School, Cambridge University.

1. Introduction: genetics of PWS

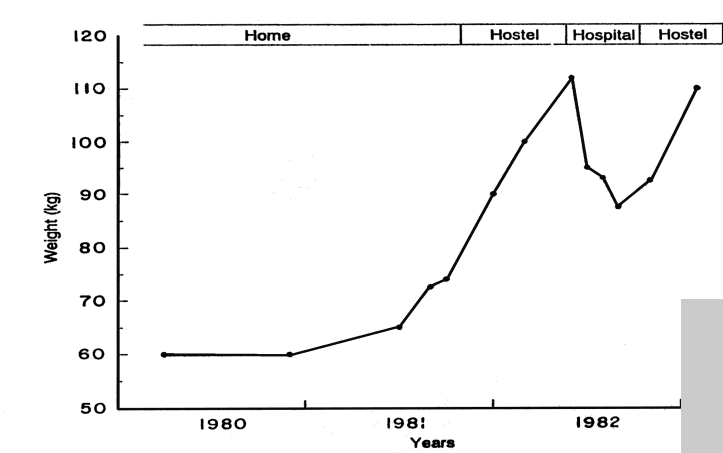
Aim: to understand the uniqueness of PWS from genes to brain to behaviour to support

Absence of expression of gene(s) imprinted (switch off on the chromosome of maternal origin), or deleted (from the chromosome of paternal origin) located on chromosome 15 at the locus q11-13.

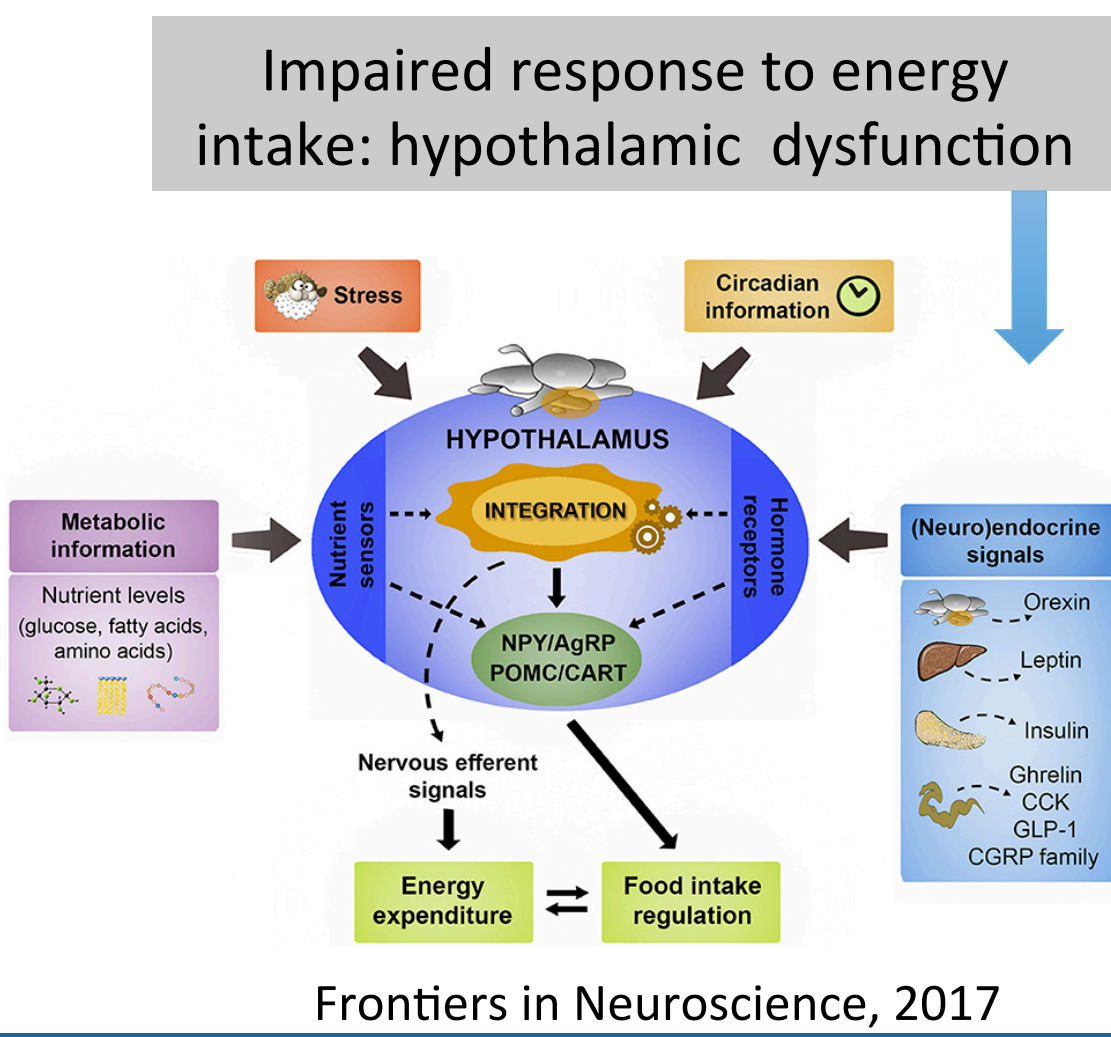
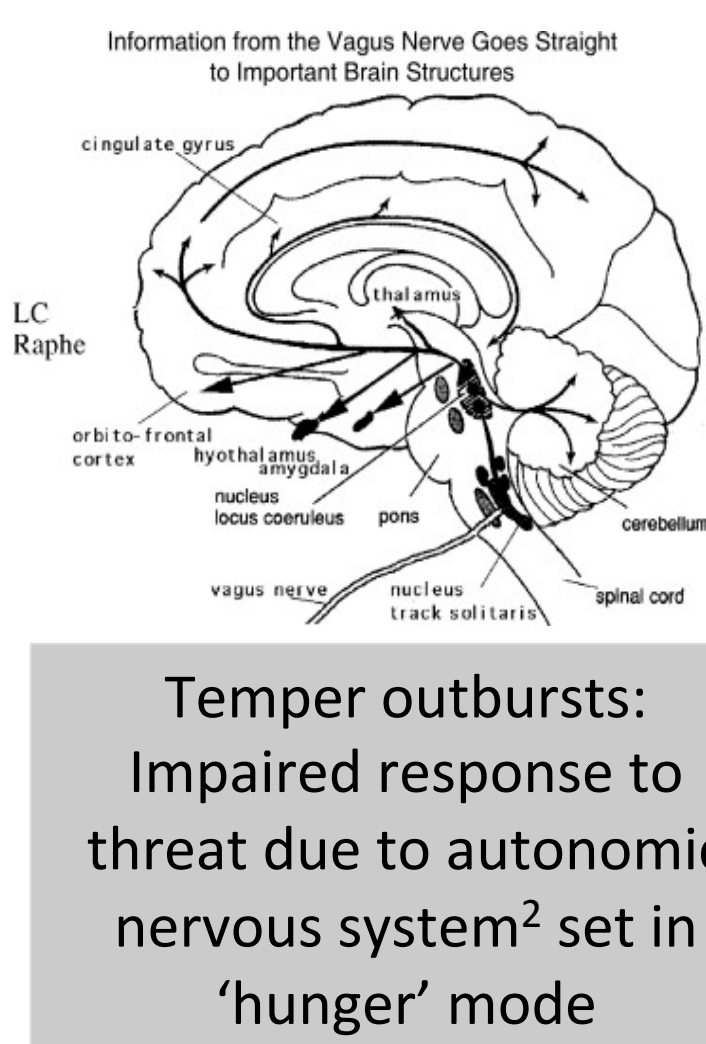


4. From genes to mechanisms to understanding

Prader-Willi Syndrome: a disorder of homeostatic regulation in a changing environment



- Energy balance and growth
- Sexual development and reproduction
- Social functioning



2. Introduction: the phenotype of PWS¹



At birth

- Extreme floppiness (hypotonia)
- Failure to thrive
- Needs assistance to feed
- Impaired sexual development (e.g. undescended testes in boys)



From early childhood

- Developmental delay
- Intellectual and social impairments
- Short stature
- Impaired sexual development
- Onset of severe over-eating (hyperphagia)
- Temper outbursts
- Repetitive and ritualistic behaviours
- Skin-picking
- Shortened life expectancy (<40 years)
- Obesity-related morbidity and mortality (e.g. diabetes mellitus, sleep apnoea)
- Hyperphagia-related morbidity and mortality (e.g. aspiration, choking)

3. Informed and integrated intervention



5. Conclusion: the challenge of support

Balancing respect for autonomy & personal choice with risk of harm (UN CPRD)

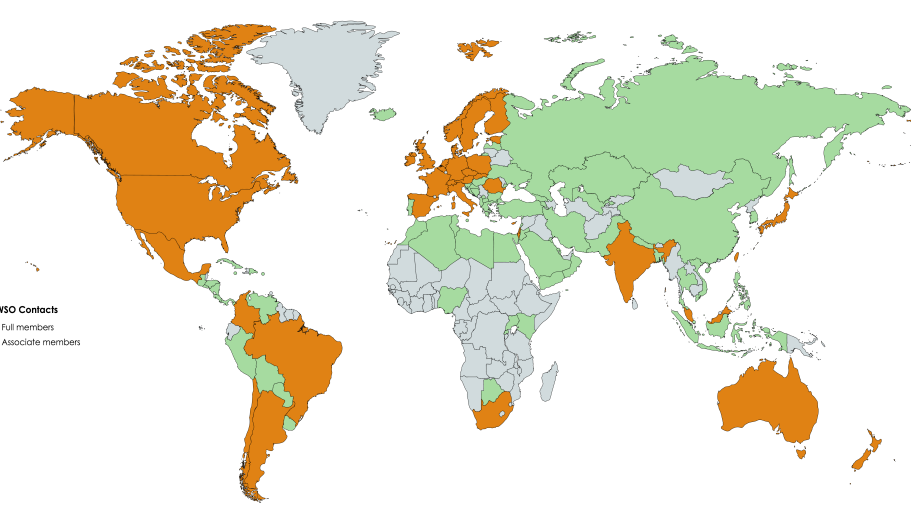
- Life-threatening obesity (e.g. refusing limitations on food)
- Behaviours puts a person with PWS at risk (e.g. exploitation, sexual and physical abuse)

Some key ethical and legal principles

- The right to, and importance of, understanding PWS
- Consent to intervention
- Proportionate response to risk of harm
- Access to informed support in the community

6. About IPWSO

International PWS Organisation (IPWSO), founded in 1991
Registered as a Charity in England and Wales
Membership 38 National PWS Associations. Contacts in over 100 countries.



www.ipwso.org
Thank you to the Board of Trustees and all who work and volunteer for IPWSO

References

- ¹Prader-Willi Syndrome. Editor: C. Höybye, Nova Medicine and Health 2013
- ²The Polyvagal theory: neurophysiological foundations of emotions, attachments, communications and self-regulation. Stephen Porges