Improving mental health and wellbeing for people with Prader-Willi Syndrome

Tony Holland, Department of Psychiatry, University of Cambridge, UK
President, International Prader-Willi Syndrome Organisation

PWSA USA Family Conference 23rd to 24th June 2023

Declarations: Advisor to pharmaceutical companies on clinical trials
World Health Organisation (WHO) definition of mental health
(Galdeseri et al. 2015, Towards a definition of mental health. World Psychiatry, DOI:10.1002/wps.20231)

Good mental health is more than ‘the absence of mental disorder’. It includes the presence of:

- A dynamic internal state or equilibrium
- The ability to maintain harmony in line with universal values
- The ability to recognise, express and modulate emotions
- The ability to cope with adverse events and function socially

This definition is analogous to that for physical health
Quality of life, wellbeing and mental health

Determinants of quality of life, wellbeing and mental health include: societal, psychological and biological factors.

Individual and shared experiences

Presence of positive attributes: contentment, a sense of worth
Improving mental health, wellbeing, quality of life
Impact of the individual, support network and society

The person with PWS

- Strengths and weaknesses
- Extent and nature of behaviours of concern
- Physical and mental health
- Their understanding & acceptance of PWS
- Access to professional expertise and treatments

Family and close support network

- Early diagnosis
- Access to information on PWS
- Family, friends and professional support
- Financial resources
- Housing

Society

- Opportunities (education, employment, leisure)
- Social inclusion
- Financial support (benefit systems)
- Civil society (PWSA, Social networks etc)
- Policies and practices that are inclusive
- Public acceptance and understanding
Good mental health

For people with PWS having and maintaining good mental health, and a sense of positive wellbeing and quality of life are not just issues for health services.

They require a range of actions and activities including from wider society and support services, access to funding and to informed health support from the relevant disciplines, and the availability of guidance and advice as and when it is needed.
<table>
<thead>
<tr>
<th>Legislation matters: changes in England and Wales</th>
</tr>
</thead>
<tbody>
<tr>
<td>How Government policies determine attitudes and the agenda</td>
</tr>
</tbody>
</table>

The Mental Deficiency Act 1913
Better Services for the Mentally Handicapped, 1971

<table>
<thead>
<tr>
<th>Equality Act 2010</th>
</tr>
</thead>
<tbody>
<tr>
<td>Making ‘reasonable adjustments’ for people with disabilities. Adapting work places, provision of aids etc</td>
</tr>
</tbody>
</table>

NHS and Community Care Act 1995

UN Convention on the Rights of Persons with Disabilities, (UNCRPD) 2006
Interrelationships between those supporting and those being supported

Families of children with Prader–Willi syndrome: stress–support and relations to child characteristics

R M Hodapp, E M Dykens, L L Masino

Affiliations

PMD: 9018579 DOI: 10.1023/a:1025865004299

• Families of children with PWS experience higher stress levels
• Families turn to family and friends for support
• Behaviour problems associated with high parental stress

Behaviours of concern and mental ill-health arise in the context of an inter-relationship between biological vulnerability and environmental factors - helping families maintain their resilience will help the mental health of those they support
Supporting families and paid care staff to meet the mental health and behaviour needs of people with PWS

What helps?

- Being prepared for different phases (diagnosis, onset of hyperphagia, the nature of PWS, planning for the future)
- Access to informal and professional support
- Structured care plans
- Strategies to prevent and manage behaviours of concern, including crisis planning

AAIDD support model
The individual with PWS: behaviour and mental health

- Atypical brain development
  - Genetic sub-type differences
  - Cortical & subcortical structures
  - Hypothalamic neural and neuroendocrine pathways

- Dimensions of mental ill-health associated with PWS
  - Abnormalities of mental state
    - Mood instability
    - Situational anxiety
    - Affective disorder
    - Psychotic illness (mUPD>deletion)

- Behaviours of concern
  - Hyperphagia
  - Emotional (temper) outbursts
  - Body-focused self-harming behaviours (e.g. skin picking etc.)
  - Repetitive & ritualistic behaviours
  - Rigidity and perseveration

- Cognition, Communication & Social Functioning
  - Impairments in intellectual & executive functioning, social cognition & communication
  - Cognitive inflexibility/rigidity
  - Intellectual Developmental Disabilities (IDD)
  - Autistic Spectrum Conditions (ASC)
Mental ill-health, including behaviours of concern, arise as a consequence of:

a) Dysfunction of the hypothalamus and its networks
   • Hyperphagia
   • Mood regulation
   • Emotional control

b) impaired brain development affecting cortical and sub-cortical regions and networks:
   • impaired cognition (incl. social)
   • reduced ability to understand and to effectively respond to change

Interaction between biological vulnerability and environmental
Mental health at time of transition to adult life

Maintaining ‘homeostasis’ in a changing world

In adult life increasing freedoms, choices, and risks

Importance of acceptance, support and opportunity

Individual characteristics of people with PWS that may impact transition
  • Cognition (higher cognitive functioning, including social cognition)
  • Hyperphagia
  • Emotional regulation and risk of outbursts
  • Risk of mental ill-health

Changes in the environment that may impact transition to adult life
  • Structured to unstructured
  • Access to food
  • Certainty (familiarity) to uncertainty
  • Increasing choice and independence
  • Level of support
Preventing and managing emotional outbursts

- Impaired perception and cognitive functioning
- Environmental change
- Increased cognitive load

Diagrams courtesy of Janice Forster

The Autonomic Nervous System (ANS)

Development and maintenance of behaviours

- Motivation
- Triggers
- Skills deficits
- Behaviours of concern
- Consequences

Environmental factors

Interventions
- Environmental
- Psychological
- Biological

Extract from diagram courtesy of Kasey Badart and Annette Griffith
Prevention and management of outbursts

Optimise sensory inputs and reduce uncertainty (anxiousness)
- food security,
- environmental support,
- visual timetables, calendars etc.
- allow time for cognitive processing
- use of language, written/visual material

Improve emotional regulation thereby improving perception and reasoning
- Training for people with PWS to manage change etc.
- Consistent agreed approach to potential triggers and when an outburst is beginning
- vagus nerve stimulation
- medication
Mental illness in people with PWS

- Risk of mental illness developing is increased in teens in people with PWS due to mUPD
- Mental illness can be treated and is an indication for the careful prescribing of psychiatric medication
- Where changes in behaviour and mental state are out of the ordinary and bizarre – asked why – importance of an accurate diagnosis and formulation

Presents with a deterioration in behaviour and/or the onset of new bizarre behaviour

Onset usually acute but can also be gradual

Associated with abnormal mood state and the development of abnormal mental experiences (confusion, hallucinations, delusions)

Interventions
  - Medication based on accurate diagnosis
  - Reduce demands
  - Consistent informed support
  - Prevention of harm
Maintaining good mental health, wellbeing and quality of life for the person with PWS

Society
- Policies, practices and attitudes that are understanding and facilitate social inclusion
  - Availability of services to meet needs

The person with PWS
- Early intervention
- Informed support
- Skills development
- Access to informed expertise and interventions

Family and professional support
- Information and understanding
- Network of support
- Finance and housing
THANK YOU

www.pwsausa.org

Info@pwsausa.org

(941) 312-0400