

ITHACA Board Meeting

2020 December 10 – 12

Satellite Meetings : Friday Dec. 11th

WG 9 ID

Sylvia Huisman PIMD SNW

Good practice recommendations for the
management of children with
profound intellectual impairment and multiple
disabilities



WG 9 ID - Launch 3 projects for the next 2 years

Diagnosis and management of intellectual disability



Discussion

- ➔ Propose 3 separate working groups
- ➔ Identify potential partners ,
 - need experts out of the genetic circle of ITHACA
 - including in the other ERNs (Epicare, NMD...)
- ➔ Provisional roadmap
- ➔ Transversal recommendation could be based on Raoul's experience (WG4) and on the means proposed by the European community (help in writing and organizing meetings.)
- ➔ **Developing a specific dataset for ID patients in ILIAD**

Recommendations of good practice for the management of mentally disabled adults

Good practice recommendations for the management of children with profound intellectual impairment and multiple disabilities

Launch oct 20
Sylvia Huisman

Recommendations of good practice on strategies for diagnosing intellectual disability, applicable in Europe



Good practice recommendations for the management of children with profound intellectual impairment and multiple disabilities

- **Focusing on genetic causes - but these recommendations could be relevant to all patients with profound, severe and multiple disabilities**
- Set a subnetwork group (SNW) on profound intellectual and multiple disabilities
- Based on the PNDS Polyhandicap (French National Diagnostic and Care Protocols (NDCPs) on PIMD profound intellectual and multiple disabilities) published in may 2020 – English Translation is in progress
- To Agree on a EU specific definition
- To Edit practical/recommendation (specific management fact sheets)



october 20

- Identify needs and partners

January 20

- Draft production and SNW meeting

July 21

- 1st production & revision

January 22

- Final proposition to be discuss

June 22

- Final publication dissemination



Group leader - Sylvia Huisman, ID physician

ITHACA PM contact - Anne Hugon

Project PIMD / produce Good practice recommendations, Transversal recommendations

- Good practice recommendations for health care of children with profound intellectual and multiple disabilities
- Good practice recommendations for daily care and support of children with profound intellectual and multiple disabilities.

- Starting from rare (genetic) causes, directing towards generic recommendations relevant to all individuals with PIMD
- Medical, rehabilitative, and socio-educational care
- To guide professional care in European countries and to empower families to receive optimal care



Goals

- Identifying specific group of people based on their characteristics (directly or indirectly related to cerebral functioning) and needs
- Holistic approach with balanced focus on physical and mental (emotional) health and well-being
- Optimal support of development and quality of life of individuals with PIMD and the ones whom they closely relate to

Points of attention

- Early signs, (proactive) care and treatment options
- Developmentally appropriate approach, communication and (by proxy) decision making
- Multidisciplinary collaboration
- Goal attainment and life path dedicated care
- Organization of continuous care, coordination and longitudinal follow-up



Target audience

- Medical, rehabilitative, and socio-educational care professionals
- Lay version for parents and daily caregivers

Cover the Period from Birth to 25 yrs

- argument to include transition period > Good practice recommendations for children and adolescents with PIMD.



Working group and task assignment

- 10 persons from various disciplines and 2 ePAG members/patient advocates
- At least 4, 5 countries FR, NL, DE, ES or IT to agree on a EU specific definition/position
- Invite 1 expert representative per country/HCP
- Collaborate with Patient Organisation
- Rely on professional and specialist' societies (ex: EPNS The European Paediatric Neurology Society <https://www.epns.info>)

Methods, immediate actions

- Collecting of existing recommendations and national guidelines, establish a consensus and propose a definition
- A minimum of consensus with 4 countries?



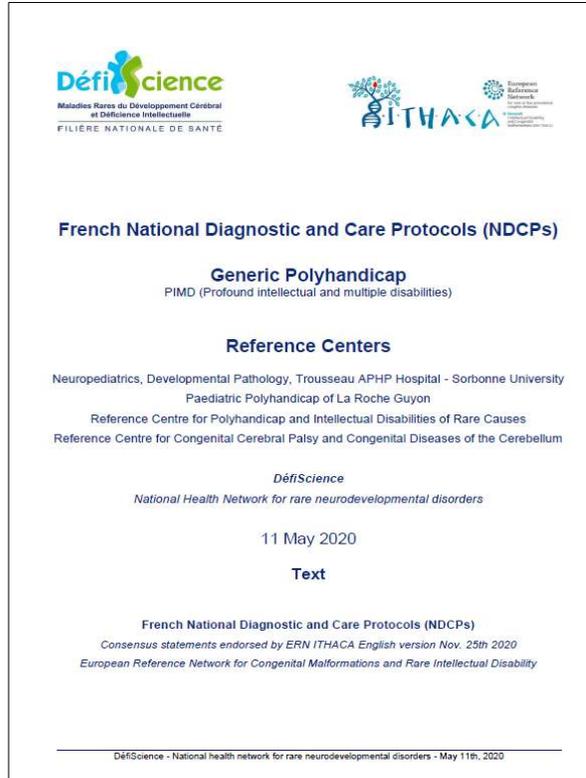
Prepare Draft Project

Tree format, online/web-based version?

Send to all HCP, with a survey

- What parts are or are not applicable in your country?
- What is missing?
- What needs to be changed?
- What needs to be added?

French guidelines, translated engl nov 2020 National Diagnostic and Care Protocols (NDCPs)



Pre-existing guidelines

- Valid sources to build on abstract,
- These guidelines largely integrated and implemented the same concepts and explanations
- **Define a general definition/classification and common**
- Terminology of this specific group of people
- PIMD term and general definition
- ≠ except for 'the signs of autism'
 - Netherlands sign of autism not used in this context
 - Italy, Poland use of behaviour
 - Roumania quite similar but no official definition

Dutch guidelines, nl 2020



What is the situation in other countries ?

We did not receive any other National guidelines
What do you think ?

<https://www.nvk.nl/>
<https://vsop.nl/>

The Home of Polyhandicap /PIMD

(starting from rare (genetic) causes)

Rare diseases in the scope of ITHACA: PIMD / Polyhandicap

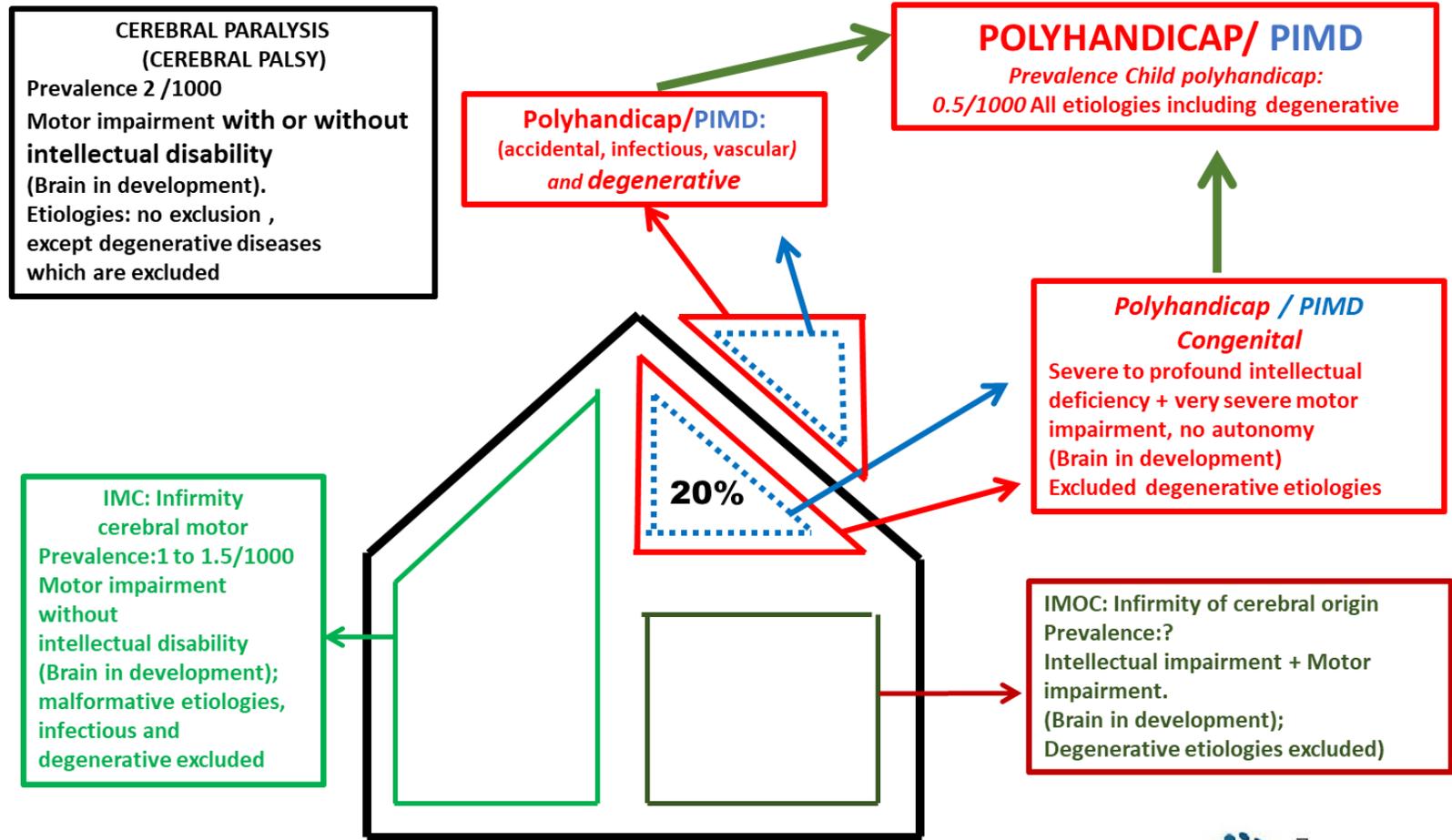
The prevalence of polyhandicap is 0.50 per 1,000, so it is not uncommon. However, the etiologies of polyhandicap are by and large rare diseases, diagnosed or not. Multiple brain lesions and their consequences on growth and secondary repercussions on various organs are sufficiently characteristic for this situation of severe disability, from birth or early childhood, to be declared as a true pathological entity in a national summary document.

Problem of linguistic and terminology

PIMD Profound intellectual and multiple disabilities



Collect EU definition from each countries, taking the basic point from the French guideline definition



The place of polyhandicap among neurological disabilities with motor impairment

Pr Gérard Ponçot

SNW2 PMID - A starting point



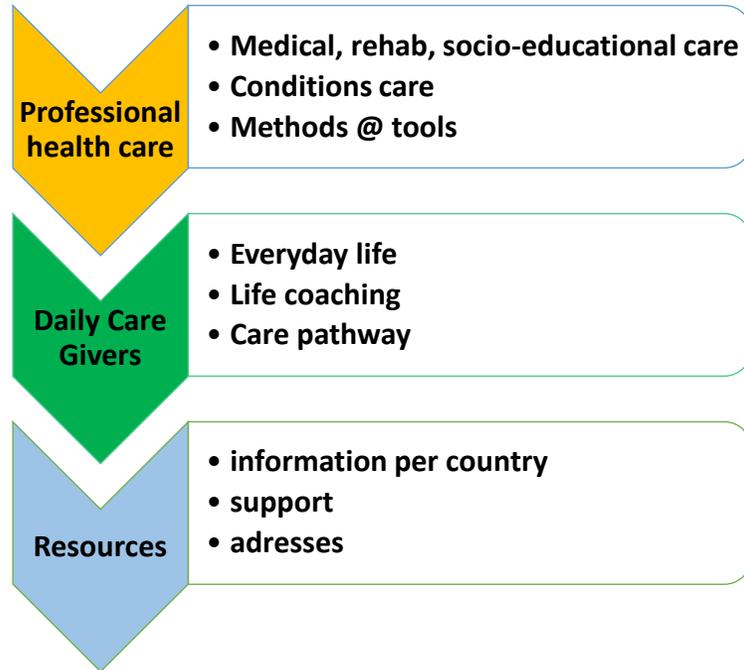
The official French definition of polyhandicap has been updated by decree N°2017-982 of May 9, 2017 on the nomenclature of social and medico-social establishments and services accompanying disabled or chronically ill individuals. This text was incorporated as soon as it was published in the Code of Social Action and Families and describes the population concerned:

- "Individuals with an early or developmental brain dysfunction resulting in severe, multiple, and progressive disturbances in motor, perceptual, and cognitive capacities and the construction of relationships with the physical and human environment, and a situation of extreme physical, mental, and social vulnerability in which some individuals may present, either temporarily or permanently, signs of autism".
- This official recognition of severe and multiple disabilities **(lack of independent walking, no meaningful oral language, profound intellectual disability)** should make it possible to provide more appropriate responses to the needs of the people thus identified. It highlights the situation of vulnerability caused by severe and early brain injury, usually before the age of two years. Since 1989, the date of the first "official" definition, other definitions have circulated, which are intended to be more complete or more positive or to include etiological elements.
- The precise **aetiology of a polyhandicap is only known in 70 to 80% of cases:** when it is known, the cause is prenatal (65 to 80% of cases), essentially genetic, including progressive neurological diseases, perinatal (10 to 15% of cases), mostly linked to extreme prematurity, or post-natal (10 to 15% of cases).
- These brain disorders always have multiple consequences, which remain progressive throughout life: multiple neurological (somatic and intellectual), orthopaedic, digestive, respiratory, sensory, bone, and other disorders (*See Appendix 4 in connection with the definition Chapter, Table 1: Polyhandicap, medical aspects*). Behavioural disorders are frequent (30 to 40%). Pain is also a part of the daily life of these individuals. The perceptive, sensitive, and affective capacities of these people, their skills, and their appetite for communication must be considered to optimize the care they receive and their quality of life.



**Collect EU definition from each countries, taking the basic point from the French guideline definition
Propose a first definition arranged in the steering group and a starting classification**

SNW2 PMID – Classification tree proposition



Professional Health Care

| Medical, rehabilitative, and socio-educational care | Conditions for optimal care and follow-up | Methods and tools |
|---|---|--|
| <ul style="list-style-type: none"> • Etiologies and diagnosis • Clinical signs and treatment • Cognitive, communicative, motor functioning • Development, behaviour and quality of life | <ul style="list-style-type: none"> • Developmentally appropriate approach, communication and (by proxy) decision making • Multidisciplinary collaboration • Goal attainment and life path dedicated care • Organization of continuous care, coordination and longitudinal follow-up | <ul style="list-style-type: none"> • Classification systems • Pain assessment tools • Behavioral assessment tools • Communications tools • Neuro-orthopaedic devices • Tools to support hospitalisations |

Daily Care Givers (layman version)

| Everyday life | Life coaching (Organisation life path) | Care pathway (patient journey) |
|---|--|---|
| <ul style="list-style-type: none"> • Specific recommendations • Therapeutic education and the promotion of health • Life path and support • Health decision making • Participation and quality of life | <ul style="list-style-type: none"> • Early childhood (0 to 6 years) • Childhood and adolescence (7 to 12 years old) • The transition from childhood to adulthood (13 to 20 years old) | <ul style="list-style-type: none"> • Information transmission and networking • Family support • Team of professionals and support staff Professionals • Health networks, reference centres, and other organizations |

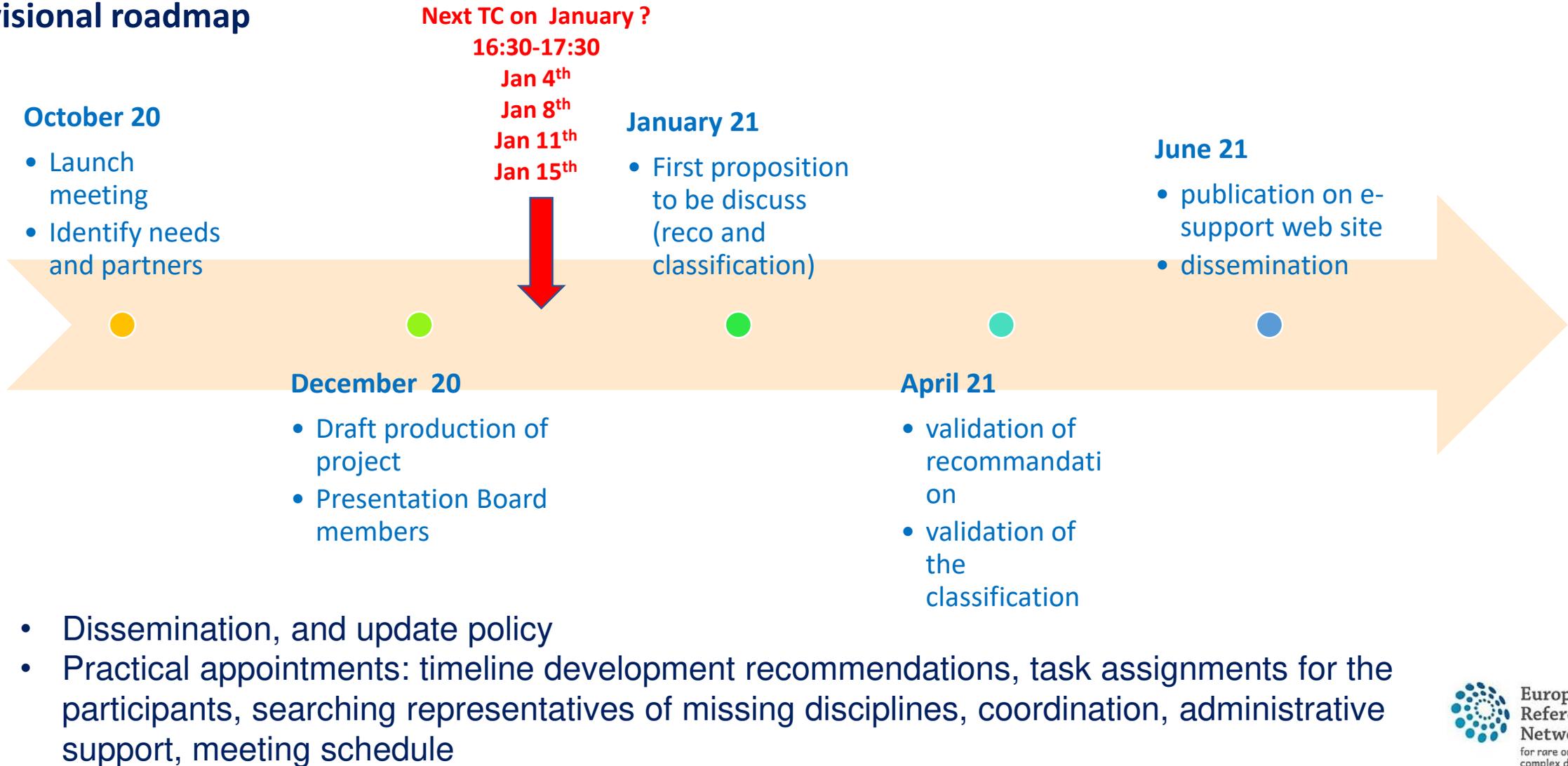
Resources international and per country

| | | |
|-----------------------------|-----------|-----------|
| • information (publication) | • support | addresses |
|-----------------------------|-----------|-----------|

SNW2 PMID – Discussion & wrap up



Provisional roadmap



Project Manager Contact: anne.hugon@aphp.fr