European Reference Network
for rare or low prevalence complex diseases

Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

NHS Manchester University NHS Foundation Trust

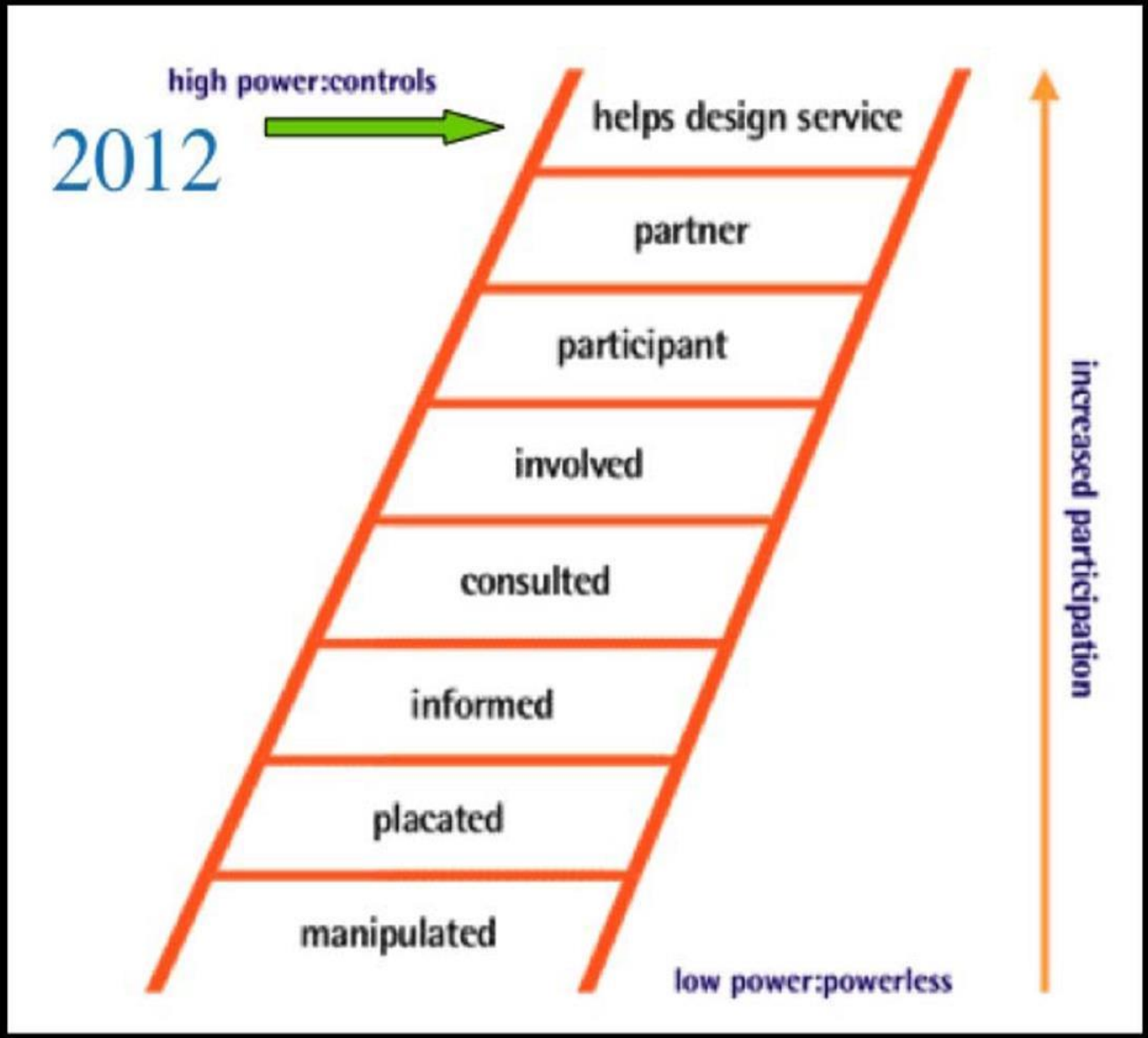
ERN ITHACA: RO-NMCA ID participation (NoRo)

Dorica Dan – NoRo

ITHACA

https://www.euractiv.com/section/health-consumers/infographic/rare-diseases-in-the-eu/

We changed the role of the patients



DIRECTIVA 2011/24/UE A PARLAMENTULUI EUROPEAN SI A CONSILIULUI din 9 martie 2011 privind aplicarea drepturilor pacientilor în cadrul asistentei medicale transfrontaliere

Articolul (54)

Comisia ar trebui să sprijine dezvoltarea continuă a retelelor europene de referință (RER) între furnizorii de servicii medicale si centrele de expertiză din statele membre.

Retelele europene de referință pot îmbunătăti accesul la diagnostic si furnizarea unei asistente medicale de înaltă calitate tuturor pacientilor a căror situatie medicală necesită o concentrare deosebită de resurse si expertiză si ar putea, de asemenea, constitui puncte centrale pentru formarea si cercetarea medicală, diseminarea informatiilor si evaluare, ales în cazul bolilor rare. Prin urmare, prezenta directivă ar trebui să ofere stimulente statelor membre pentru a consolida dezvoltarea continuă a retelelor europene de referință.

Retelele europene de referință se bazează pe participarea **voluntară** a propriilor membri, dar Comisia ar trebui să formuleze criterii si conditii pe care retelele ar trebui să fie solicitate să le îndeplinească pentru a primi sprijin din partea Comisiei.

RETELELE EUROPENE DE REFERINTĂ (ERNs)

SCOP:

Îmbunătățirea accesului la îngrijiri medicale ultraspecializate, de înaltă calitate și sigure.

POLItica de sănătate europeană pentru bolile rare- ACTIVITĂȚI ale comisiei

1. Planuri și strategii în domeniul bolilor rare:

EUROPLAN:

Recomandări pentru elaborarea planurilor și strategiilor naționale privind BR (2008-2011) și (2012 – 2015).

2.Definirea, codificarea și inventarierea bolilor rare

3. Cercetarea cu privire la bolile rare

Platformă europeană privind înregistrarea BR.

4. Centre de expertiză și rețele europene de referință în domeniul BR

SURSA: Raport privind punerea în aplicare a Comunicării Comisiei intitulate „Bolile rare: o provocare pentru Europa” [COM(2008) 679 final] și a Recomandării Consiliului din 8 iunie 2009 privind o acţiune în domeniul bolilor rare (2009/C 151/02), 2014

POLItica de sănătate europeană pentru bolile rare- ACTIVITĂȚI ale comisiei

5.Reunirea expertizei în domeniul bolilor rare la nivel european

6.Responsabilizarea organizațiilor de pacienți

7.Guvernanța și coordonarea europeană

8. Acțiuni de ameliorare a calității asistenței medicale în domeniul BR → Screeningul populației pt depistarea BR → Facilitarea accesului la produse medicamentoase orfane → Regulamentul privind produsele medicamentoase orfane

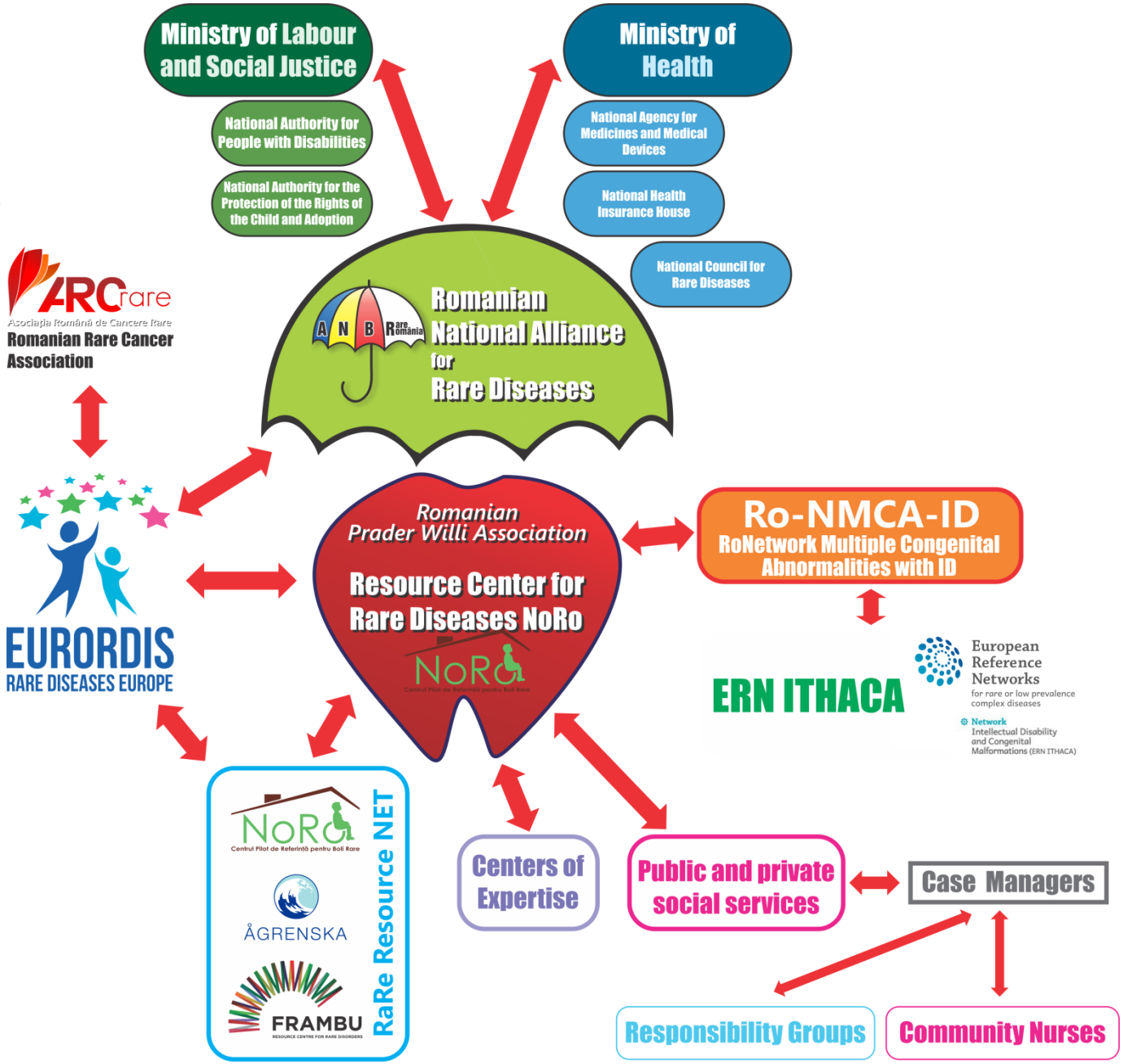
9. Dimensiunea globală a politicii în domeniul BR

SURSA: Raport privind punerea în aplicare a Comunicării Comisiei intitulate „Bolile rare: o provocare pentru Europa” [COM(2008) 679 final] și a Recomandării Consiliului din 8 iunie 2009 privind o acţiune în domeniul bolilor rare (2009/C 151/02), 2014

RECOMANDĂRILE COMISIEI EUROPENE:

UNDE SUNTEM?

CUM NE RAPORTAM LA CEILALTI?



EURORDIS
RARE DISEASES EUROPE

The network

Ro-NMCA-ID (RoNetwork Multiple Congenital Abnormalities with ID):

- 1. L.Turcanu Paediatric Emergency Hospital **Timisoara** (Regional Centre for Medical Genetics Timis);
- 2. NoRo Pilot Reference Centre for Rare Diseases in **Zalau**;
- 3. County Emergency Hospital **Craiova** (Regional Centre for Medical Genetics Dolj)
- 4. “Sfanta Maria” Paediatric Emergency Hospital **Iasi**, Medical Genetics Center
- 5. Municipal Hospital ”Dr. Gavril Curteanu” **Oradea** (Regional Centre for Medical Genetics Bihor)


ROMANIA
National capital
County seat
City, Town
Major airport
International boundary
Expressway
Main road
Railroad
SLOVAKIA
UKRAINE
HUNGARY
SERBIA
BULGARIA
MOLDOVA
UKRAINE
Chernivtsi
Debrecen
Békéscsaba
Oradea
Zalău
Satu Mare
Carei
Baia Mare
Sighetu Marmației
Viseu de Sus
Borșa
Tășnad
Jibou
Dej
Bistrița
Vatra Dornei
Suceava
Siret
Dorohoi
Botoșani
Fălticeni
Hârlău
Pașcani
Iași
Bălți
Chisinau
Odessa
Piatra Neamț
Roman
Toplița
Salonta
Cluj-Napoca
Beius
Cermei
Ștei
Ineu
Turda
Târgu Mureș
Sovata
Gheorgheni
Bacău
Vaslui
Huși
Miercurea Ciuc
Odorheiu
Arad
Abrud
Brad
Blaj
Zlatna
Alba Iulia
Deva
Hunedoara
Cugir
Sibiu
Târgu Secuiesc
Covasna
Sfântu Gheorghe
Tecuci
Timișoara
Lugoj
Hateg
Râșnov
Brașov
Focșani
Moldoveanu Peak
2,544 m (8,346 ft)
Galați
Caransebeș
Brezoi
Sinaia
Râmnicu Sărat
Brăila
Reșița
Anina
Târgu Jiu
Râmnicu Vâlcea
Buzău
Tulcea
Sulina
Ploiești
Belgrade
Pitești
Babadag
Drobeta-Turnu Severin
Orsova
Găești
Pogoanele
Urziceni
Slobozia
Tândarei
Craiova
Slatina
Bucharest
Fetești
Medgidia
Călărași
Constanța
Black Sea
Rosiori de Vede
Oltenița
Alexandria
Silistra
Giurgiu
Turnu Măgurele
Ruse
Mangalia
Dobrich
Kavarna
Pleven
Danube
0 25 50 75 100 km
0 25 50 mi
N

Colaborarea Ro-NMCA-ID

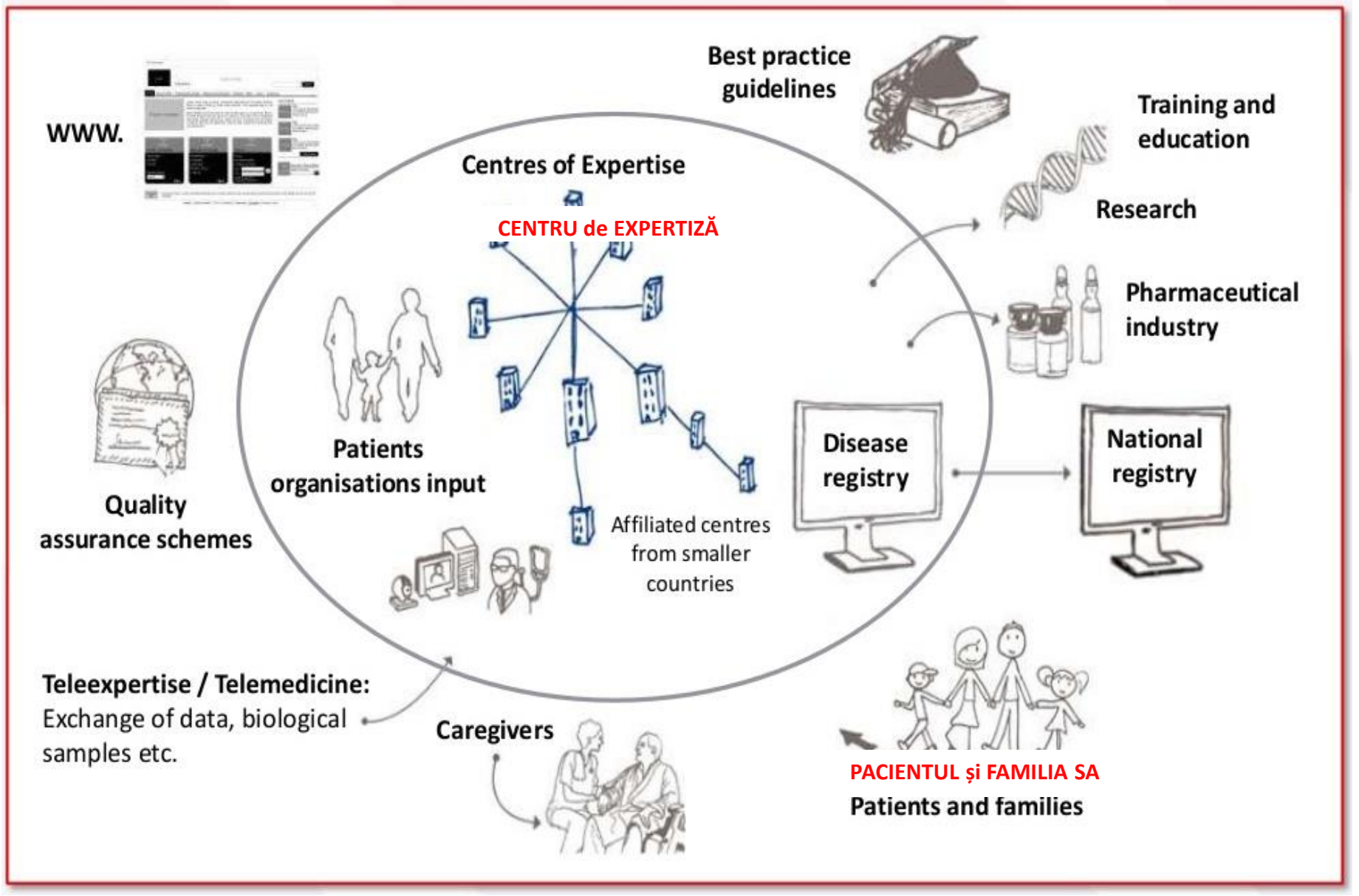
Ro-NMCA-ID se bazeaza pe o activitate de colaborare de peste 5 ani inainte de formarea retelei

Cele 4 centre de genetica au expertiza in diagnostic, prevenire si tratament in bolile genetice, in timp ce NoRo ofera terapie, educatie terapeutica si grupuri de suport;

- ✓ *"The Centre in **Timisoara** brings a rich **experience with paediatric patients** and high throughput **array CGH** and **sequencing equipment**.*
- ✓ *The Centre of in **Craiova** brings expertise in **prenatal** testing and prevention of rare genetic diseases.*
- ✓ *The Genetics Departments from **Iasi** and **Oradea** have broad expertise in **dysmorphology**.*
- ✓ *The **NoRo Centre** completes the network with **supportive medical and social services, therapies, patient/family empowerment and trainings**, alongside efforts to **raise awareness for rare diseases.** "*

Etape	Obiective	Responsabili NoRo	Infrastructura
1	Pacientul se adreseaza Centrului NoRo pentru diagnostic (HelpLine) _ Este orientat pentru diagnosticare la unul din centrele de expertiza cu care colaboram;	Operator HelpLine	HelpLine NoRo
2. 1.	Diagnosticare intr-unul din centrele din retea	Specialisti CE	Catedra de Genetica Cluj, Centrul de Genomica sau CRG Tm, Iasi, Oradea, CRGD, IOMC, CE, Clinica de Reumatologie Cj, CETM, CE Fundeni, CE Parhon, etc.
2.2.	Stabilirea unui diagnostic clinic corect cu ajutorul testelor specifice aflate in dotarea NoRo ;	Genetician pediatru Medic specilist psihiatrie pediatrică Psiholog clinician Neurolog	NoRo (LMD, Ecograf, EKG, EEG, EMG, Extractor ADN, teste psihologice)
3	Consultul clinic si genetic al pacientilor diagnosticati; Daca diagnosticul nu este de natura genetica, se trece DIRECT la pasul 6;	Genetician Pediatru NoRo Psihiatru NoRo Neurolog NoRo	Cariotip si FISH – testarea la Catedra de Genetica, UMF Cluj, Centrul de Genomica Tm, CRGD, CRGI; Alti specialisti din CE
3	Transmiterea analizelor pentru efectuarea testelor genetice specifice la cei cu tablou clinic sugestiv pentru o anomalie genetica;	Genetician Pediatru NoRo	Cariotip si FISH – testarea la Catedra de Genetica, UMF Cluj, Tm, Centrul de Genomica Tm, CRGD;
4	Analiza globala a genomului prin SNP array sau cariotip, daca este cazul;	Genetician Pediatru angajat al Centrului NoRo	Catedra de Genetica Cluj, UMF Timisoara, CRGD;
5	Acordarea sfatului genetic post- testare pentru pacientii diagnosticati	Genetician Pediatru NoRo Psiholog NoRo Reprezentant pacienti	Grup de suport Centrul NoRo Consiliere psihologica si informare privind serviciile accesibile (NoRo)
6	Initierea conduitei terapeutice si a tratamentului (daca este cazul) ;	Specialist CE	Catedra de Genetica Cj, UMF Timisoara, CRGD; CRGO, CRGI, IOMC, TM, Pediatrie 1, Clinica de Reumatologie Cluj, Clinica Fundeni, Inst. Parhon. Alte CE;
7	Introducerea in serviciile Centrului NoRo	Echipa interdisciplinara NoRo	Infrastructura de educatie terapeutica, respiro si recuperare: Ambulator NoRo Centru de zi si centru rezidential,Registru de pacienti
8	Comunicarea permanenta cu celelalte servicii : CE + servicii socio-medicale la nivel local	Echipa interdisciplinara NoRo	Intalniri periodice Actualizare permanenta a hartii serviciilor Formularea de documente comune: www.participrare.ro
9	Re- evaluare	Specialisti CE	CE

Ingrijire integrata



Reprezentare Ro-NMCA-ID in ITHACA

- **Coordonator** – Centrul Regional de Genetica Timis, **Prof. Dr. Maria Puiu**
- **Membra in Boardul ITHACA, Prof. Dr. Cristina Rusu, Iasi**
- **Membra in Boardul ITHACA, Dorica Dan, reprezentant ePAG**.
- **Membra supleanta in boardul ITHACA, Dr Adela Chirita-Emandi**

European Reference Networks

Workpackages ITHACA (NoRo)

- WP1-3 Management, Dissemination, Evaluation
- WP4 Telehealth
- WP5 Expert Patient Care
- WP6 Patient Registers
- WP7 Research
- WP8 Teaching and Training

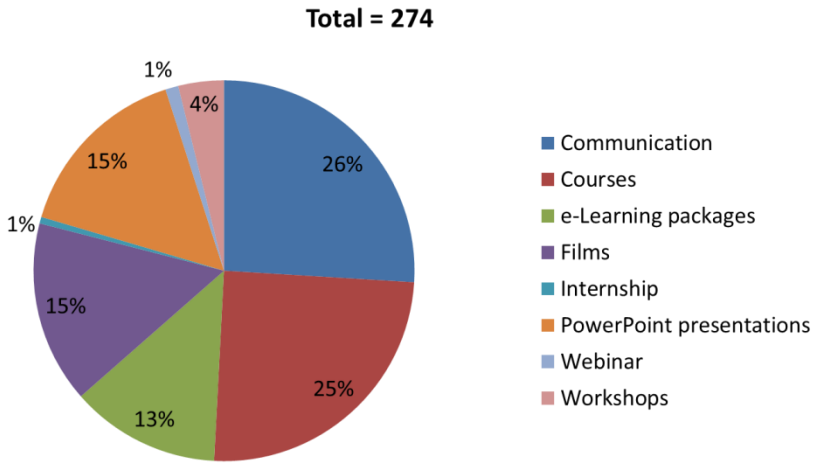
Coordinated by Manchester , transferred to Paris

- 38 centres from 14 member states
- Patient representation; E-PAGs/others
- Possibility to have new members affiliate HCPs who do not fulfil all criteria
- Involvement of other associated groups ho are not Health are Providers e.g. EUROCAT

There are currently a total of 274 materials declared. The breakdown of participation and categories of the submitted material are shown in the charts below.

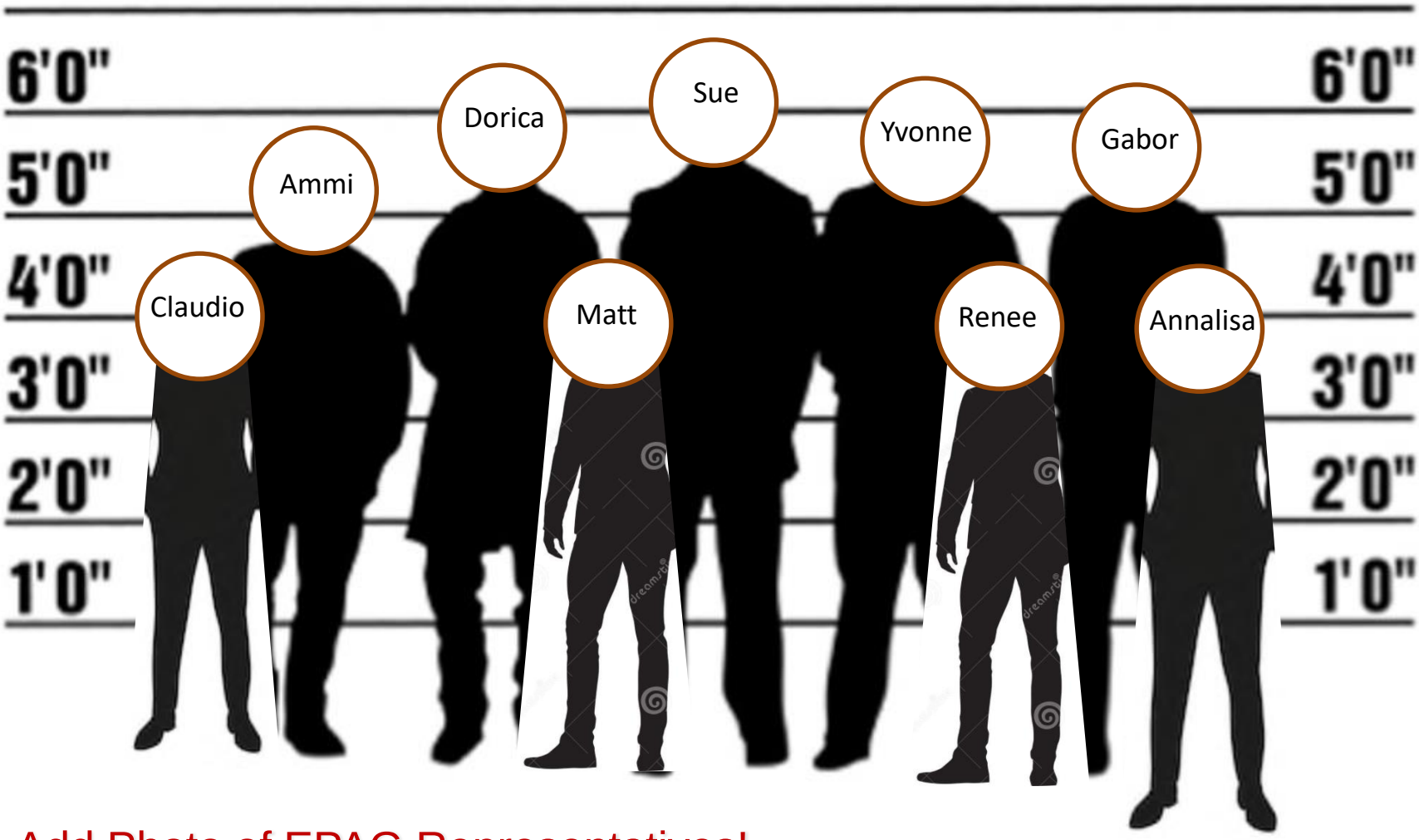
Materiale de instruire

Country	Nb HCP	Nb material	Country	Nb HCP	Nb material
Belgium	0/3	-	Italy	3/8	48
Cyprus	0/1	-	Lithuania	0/1	-
Czech Republic	0/1	-	Netherlands	2/5	17
Finland	0/1	-	Portugal	1/1	41
France	3/8	84	Romania	1/2	30
Germany	0/3	-	Sweden	0/1	-
Hungary	1/2	34	UK	1/3	20



The Usual Suspects…!

Ern-ithaca epag : Usual Suspects…!



Add Photo of EPAG Representatives!

Realizari ITHACA…

- Staff angajat (Michael Smith, Myfanwy (Miffy) Rawson)
- Rezultatele anului 1 – la timp
- Raport tehnic – trimis
- Raport financiar anul 1 - trimis
- Launch meeting Sept 2017
- Steering Group met October 2017
- Pregatirea planului de dezvoltare pentru 3-5 ani
- Aplicatii proiecte
- Intalnirea anuala la Milano
- Intalnirea F2F Paris, octombrie 2018
- Materiale instruire trimise
- Pregatire curs online
- Participare webinare, EURORDIS in Viena, EURORDIS Winter School in Paris, EU Facilitating Research meeting in Dublin, RD Action workshops
- Website ITHACA
- Webinarii trimestriale
- Modul eLearning integrated care

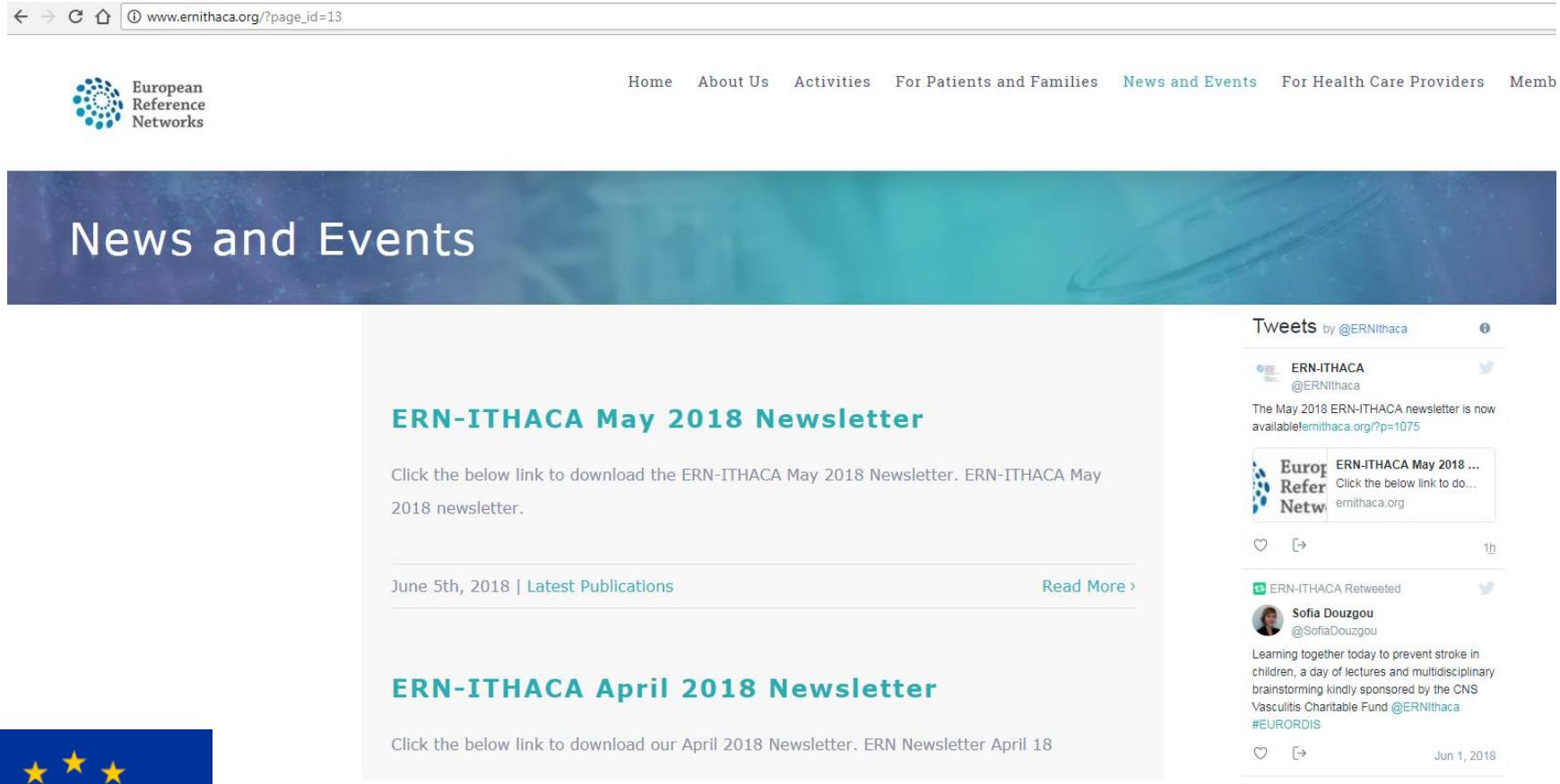
Traduceri ghiduri clinice

2 ghiduri noi: Noonan syndrome and Fetal Valproate Syndrome expert consensus

Preliminary work on next guideline with MCAP patient groups

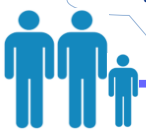
Amsterdam Pitt Hopkins guidelines being prepared

Website.... http://www.ernithaca.org/?page_id=15



www.ernithaca.org

Patient Journey Rett Syndrome



Note: Most difficult and challenging time for the family, who believed their child to be healthy!

1

… symptoms and features of the disorder become apparent

Note: Symptoms and clinical presentation become more pronounced!

2

… post diagnosis

3

… regular checks – x-rays and scans …

Note: Intense therapeutic approaches could further improve the prognosis of Rett syndrome people!

4

… referral to appropriate specialist when health need identified …

1. First Symptoms

Regression (9-30 months)
Some difficulty feeding
Floppy and poor coordination of limbs, trunk and stereotypic hand movements,
Walking, if achieved at all becomes unsteady,
Head circumference if normal at birth slows.
Onset of epileptic seizures
Abnormal breathing patterns emerge; breath holding, hyperventilation, air swallowing
Eating, chewing and swallowing problems
Heart rhythm abnormalities
Social withdrawal, confused (not autism).
Partial of complete loss of spoken language
Gastro intestinal problems

Note: Detailed care plan needs to be put in place to address the health needs of the specific comorbidities!

Ideally: Support for the family can be accessed from the experienced and professional national Rett organization (Rett UK). Access to knowledgeable and supportive GP

2. Post Diagnosis

Need for involvement of multi-disciplinary team and co-ordination of care to manage symptoms with a care plan.
Referral to paediatric (child) or neurological consultant (adult)

Ideally: Care plan to involve physiotherapy, occupational therapy, SaLT, hydrotherapy and music therapy - can help a person with RTT lead a happy and full life.
Have an up to date Health Passport for use in consultations or emergency

3. Surgery

Scoliosis
Removal of stones (gall bladder or kidney)
Other orthopaedic interventions, eg hip, feet
Gastro-intestinal surgery when symptoms become severe
Dental surgery often needed under anaesthetic

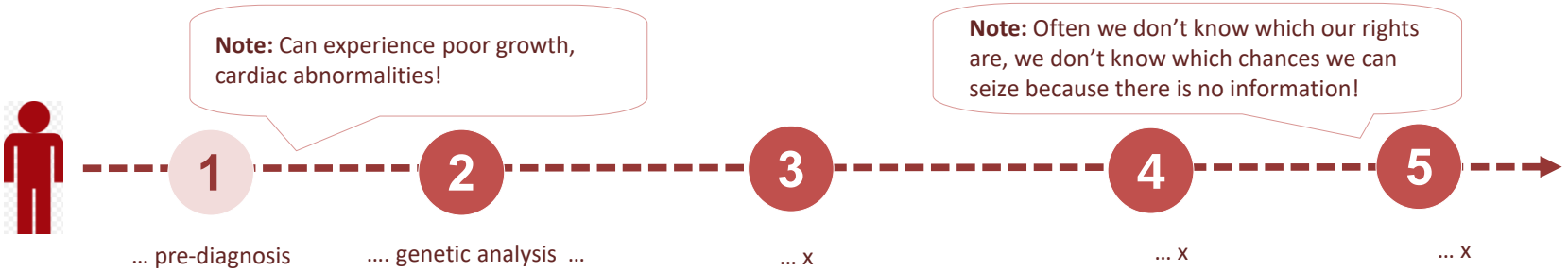
Ideally:
Regular checks including X ray or scans where appropriate to check on progression of any identified problems.

4. Follow-Up…

Problems may exist when passing from paediatric to adult health care services
Illnesses may be wrongly attributed to "having Rett syndrome," when in reality the condition is found in a neurotypical person of the same age and should therefore be treated in the same way.

Ideally: Annual health check with GP; include long QT check, blood tests, full body examination
Any symptoms identified should be promptly referred for further investigation and escalated up the care pathway where necessary
Need easy to understand guidelines/health checklists to guide parents and carers through the complex symptoms and problems of the disorder

Patient Journey Williams Syndrome



1. First symptom
Supravalvular aortic or pulmonary stenosis,
Low growth, characteristic Physiognomic aspect
Hypercalcemia
Feeding difficulties
Dental abnormalities
Hernias
Hyperacusis
Musculoskeletal problems
High urinary frequency - nocturnal enuresis

Ideally:
A centre of competence that could bring together all the symptoms asking to a geneticist to do specific analysis.
The capability to recognize symptoms and quickly give to parents a diagnosis.

2. Diagnosis & 1st Treatment
Heart controls (surgery if necessary)
Neuro - psychomotricity
Logopaedia
Physiotherapy
Music therapy

Ideally: The chance to do all the analysis in the same place;
Capability of communication to afford the first steps into the Syndrome;
Start the early intervention to accelerate the development (especially motion perception and micromanipulation.

3. Clinical & Mental Check
Clinical follow up (twice yr)
Table of growth specific for WS and guidelines about what to do at the different ages (cardiological controls, pressure, endocrinological controls, and many others)
Neurological FU (each yr) to evaluate the stage of mental development and the gap with the normal parameters

Note: Case Manager / Coordinator and Multi-Professional Approach!

Ideally:
All the therapy needed to express at the best all the potentiality of the person

4. Follow-Up
Program of mental improvement, attending to a neuro-psychotherapy x2 week until 6/8 years;
Speech therapy programme (from 2 to 4/5);
Music therapy all lifelong;
Educational program to improve their autonomy.

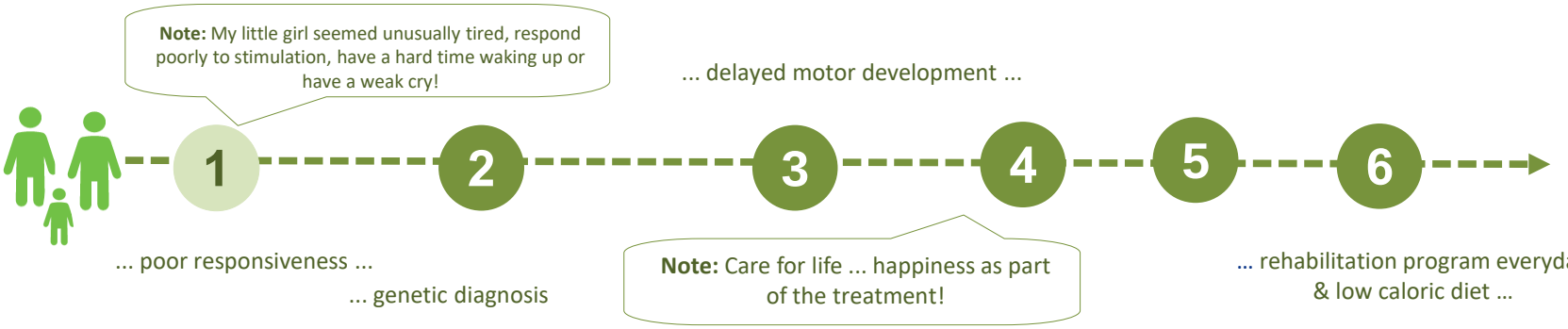
Note:
Coordination Centre to case manage a multi-professional team (teacher, therapists, doctors, family, educators....)
See MDT x twice yearly
Aim to improve capability and quality of life!

Ideally: Continuous development programme (e.g.: in a resource centre)

5. Social Care
Educational support at home to develop autonomy
Psychological support to afford anxiety and to implement awareness about "self" /who they are, which are their limits, why they can't do what other people are usually doing etc.).
Occupational therapy
Recreation programs

Ideally:
We need a well-structured social system that could give us a guideline about all the things needed. We should find answers to our questions and solution for our problems!

Patient Journey Prader Willi Syndrome



1. Pre-Diagnosis: Poor muscle tone; Distinct facial features
Poor sucking reflex and difficult feeding; Almost no cry.

1st Symptom: Hypotonia and difficulties in feeding;
Poor responsiveness; Sitting up and walking — later Hypotonia and difficulties in feeding; unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry.
Sitting up and walking later.

Ideally:
Improvement of muscle tone
Special technics for feeding and stimulating exercises
Special technics for feeding

Medical treatment and early rehabilitation intervention

3. Diagnosis
Unusual food-seeking behaviors, hypogonadism
Mild intellectual disability; Delayed motor development
Speech problems
Small hands and feet
Thick saliva and dental problems associated
Overweight

Note: Medication & rehabilitation
Support for education & school integration & inclusion into community

Ideally: GH Treatment
Sex hormone treatment
Permanent rehabilitation program; Education and work
Independent life skills

4. First Treatment
Delayed in motor development
Rehabilitation program, 3 weeks every 3 months

5. Surgery
Scoliosis
Physotherapy

Ideally:

Improvement in general health and reduced weight

6. Follow-Up Care...
Delayed in motor and intellectual development, behavior problems, Speech problems
Small hands and feet,
Overweight and sleep apnea;
Medication & rehabilitation
Support for education & school integration & inclusion into community; a proper job and qualification

Ideally: Maintaining the weight, even reducing it
High school, passing exams and training courses to find a proper job;
Integration into a Center for occupational therapy and supported job;

Patient Journey Spina Bifida

Note: Might be detected in the womb at an ultrasound control, defect on the spine or an unnatural big head.

Note: Patient will be at the hospital/clinic/specialist center for 4-7 days so it is possible to do all the exams!

… at 16-19 wks of pregnancy …

… at birth …

… diagnosis straight after birth …

… from birth to departure from life.
First time when the baby is born. After that needs a checkup every year …

1. & 2. Pre-Diagnosis & 1st Symptom
The child is born with Spina Bifida which is showed as a malformation on the back. Information about Spina Bifida from HCP and a visit from another parent with a child with SBH after approx 4 weeks after birth. Depending on how the parents are coping.

2. Diagnosis
IF the malformation is low down on the spine
= less damage

If the malformation is high up on the spine
= more damage
Measure the head to look for hydrocefalus

3. First Treatment
Surgery straight after birth within 24 hours.
Close up the back
If the patient has hydrocefalus.
Put in a shunt in the ventricle.

5. Follow-Up Care

Peadiatric Nurologist as Clinical Lead and Urologist and/or Urotherapeut,
Orthopedic,
Neurosurgeon and Radiologist

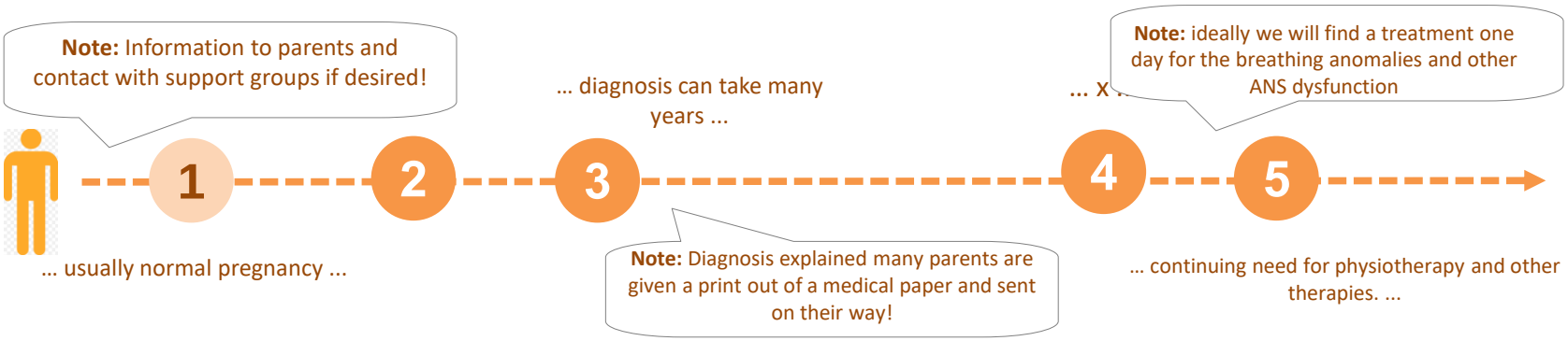
Note: *The parents can choose about proceeding with the pregnancy, surgery in the womb or leave it as it is and wait until birth!*

Ideally:
The parents feel their being well taken care of, having faith in the doctors. Being well informed on why and what to do next

Ideally:
The parents have a patient responsible doctor who has all the information on the child and who is listening to parents concerns

Note:
Urologist/urosergeon – checks up the kidney, bladder and bowl movement. If there are any problems a discussion is needed for surgery.
Urotherapist – examine bladder control and bowl movement. Teach how to do clean intermittent catheterization with is very important to be able to do yourself.
Orthopedic – checks for defects on feet's, knees, hips, scoliosis, kufosis, etc. If needed – consult with an orthopedic technician or surgery.
Neurosurgeon – Hydrocefalus, Arnold chiari syndrom, tethered cord which can do a lot of damage like lost of feel, pain and so on. If problems there is a need of surgery
The Neurologist – is the patient responsible doctor who checks out the rest like epilepsy, eyes pressure, cognition, ability to swallow and eat, weight (to big /to small) etc and writes referrals specialists in the area for further treatment.

Patient Journey Pitt Hopkins Syndrome



1. & 2. Pre-Diagnosis: Usually normal pregnancy. Some ultrasounds may show something, some may have initial feeding problems but others not. Parents will begin to onotice delay in developmental milestones, as child grows.

1st Symptom: Lack of smiling at 6 weeks and all motor milestones usually delayed. Distinct facial features which although dysmorphic not strikingly. Hypotonia.

3. Diagnosis & 1st Treatment
Blood test only available since 2007 but needs a geneticist to suspect PTHS MRI may find some structural anomalies. Distinct facial features, motor and cognitive delay, hypotonia, lack of speech, breathing regulation anomalies, unstable gait if walking, GI problemsconstipation, reflux myopia, slender fingers, palmar crease, stereotypies

4. Surgery
Some children may need foot surgery if their feet pronate too much. Some may need surgery for undescended testes. Possibly for strabismus

5. Follow-Up
Many children need medication for constipation, reflux, seizures. Some have oxygen for breathing anomalies.

Ideally:
Doctors take parents' concerns seriously early so therapy can begin promptly. Initially this is physiotherapy.

Note: Physiotherapy once doctors agree there is developmental delay. In addition children often receive OT, ST, MT and SIT. Regular follow-up by paediatrician. May have medication for constipation or reflux

Note: Sight and hearing checked. Feet looked at and reviewed regularly for specalist footwear. Back checked for scoliosis. EEG done for base-line. Advice for constipation. OT assessment for equipment

Note:
Hydrotherapy available after surgery and other physiotherapy***!***

Ideally:
Good communication between different specialists and therapists.

Note: SEN schools involvement early on. Respite for family. Help with care in home. Regular access specialist***!***

Provocari

- Start intarzaiat si timp pentru formarea echipei
- Implicarea membrilor
- Platforma CPMS
- Lipsa fonduri pentru a angaja personal
- Termene scurte pentru aplicatii
- Documente de raportare
- Timp pentru traducerea materialelor existente
- Brexit