RO- NMCA ID – Accreditation of CoE in Romania and organization of a national network: NoRo, Genetic Centers in Oradea, Timisoara, Iasi and Craiova

Presentation by Prof Dr. **Prof Maria Puiu**

Coordinator of Genetics Department in Hospital and University

Coordinator of the Centre for Medical Genetics Timis

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)

The network: **Ro-NMCA-ID**

- **Chief Executive Officer of the Healthcare Provider**
- Prof Dr **Maria Puiu**
- Tel: **0040730118152** E-mail: **maria_puiu@umft.ro**

- **Representative who will participate as a member of the Board of the Network:**
- Prof Dr **Cristina Rusu**
- Tel: **0040745432077** E-mail: **abcrusu@gmail.com**

- **Substitute representative who will participate as a member of the Board of the Network:**
- Dr **Adela Chirita-Emandi**
- Tel: **0040724369599** E-mail: **adela.chirita@umft.ro**

Existing collaborations Ro-NMCA-ID

Ro-NMCA-ID is based on five existing structures with previous collaboration.

The hospitals have expertise in diagnostic, preventive and treatment procedures in genetic diseases, while NoRo centre offers educational and supportive therapy.

- ✓ 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, patient/family empowerment**, alongside efforts to **raise awareness for rare diseases.**

Aim

- Our network aims to provide integrated services and ensure continuity of care for **people with rare congenital abnormalities and/or intellectual disability**.

- Our network is focused on a multidimensional approach to performance assessment in order to favour and to **disseminate values at national level and initiate or support quality improvement strategies**.

Area of expertise

- All four Genetic Centres provide genetic services for a broad area of genetic diseases.
- The team from **Timisoara** has gained increased expertise with **Prader Willi and Angelman Syndromes** also through research and grant projects.
- The team in **Craiova** has a strong expertise in **prenatal genetic screening** and diagnosis of chromosomal disorders, but also with **X-linked intellectual disabilities and microdeletions/microduplications syndromes testing**.

Area of expertise

- The teams in **Iasi** and **Oradea** have a broad experience in dysmorphology and a long term tradition (more than 30 years) in the diagnosis of :
- ✓ multiple congenital anomalies and intellectual disability (especially X-linked intellectual disability, subtelomeric rearrangements and microdeletions/microduplications).

- Other fields - in Iasi refer to RASopathies, ciliopathies, chromosomal syndromes, autism spectrum disorders and fetal alcohol syndrome.

Area of expertise and Range of services

NoRo was established in June 2011 as the first Pilot Reference Centre in intellectual disability (ID) from Romania, with:

- ✓ Day Care Centre for 50 children with ID and autistic spectrum disorders and Residential centre for patients with rare diseases.
- ✓ NoRo Centre provides clinical assessment and diagnosis through multidisciplinary approach (psychiatrist, neurologist, geneticist and paediatrician), psychological and social evaluation, genetic counselling, behavioural, speech, physical and sensorial therapy, hydrokinetic–therapy, ergo-therapy, weight management, educational and entertainment activities, patient registry
- ✓ NoRo is accredited for online training for patients, families, general public and professionals.

General criteria HCP – Institutions

- The Regional Centres of Medical Genetics from Timisoara, Oradea and Craiova were formed in 2014 on mature existing genetic departments in

- "Louis Turcanu" Emergency Hospital for Children Timisoara,
- County Emergency Hospital Craiova
- Municipal Hospital "Gavril Curteanu" Oradea.

- Each have five hospital beds (inpatient care), outpatient care facilities and laboratories.
- "Sfanta Maria" Paediatric Emergency Clinical Hospital Iasi includes an outpatient unit, a hospital sector with 3 beds and lab services

General criteria HCP – equipment

- The **Centre in Timisoara** collaborates with the Centre of Genomic Medicine - University of Medicine Timisoara, that includes:
- Next Generation Sequencing Lab (Illumina Technology MySeq, HiSeq2500),
- Microarray Lab (InnoScan and IScan Illumina),
- Quantitative Genotyping Lab,
- Bio IT Lab, Pre PCR Lab,
- Cytogenetic lab, Translational Research Lab
- Metabolomics and proteomic labs.

General criteria HCP – equipment

The laboratory within the **Regional Centre of Medical Genetics from Craiova** performs:

- ✓ conventional cytogenetic analysis,
- ✓ RT-PCR based methods(ViiA7-Applied Biosystems),
- ✓ QF-PCR (ABI-PRISM 3130xl sequencer);
- ✓ MLPA(CEQ8000-Beckman Coulter sequencer),
- ✓ arrayCGH/CGH + SNParray(Agilent) and
- ✓ Sanger sequencing(CEQ8000-Beckman Coulter sequencer).

General criteria HCP – equipment

”Sfanta Maria” Paediatric Emergency Clinical Hospital Iasi includes a Medical Genetics Centre since 1985.

The center has been nominated as Medical Genetics Regional Center in 2014 and is in due course to be recognized.

It covers approximately 20% of the Romanian population.

The centre lab services are provided by cooperation with the Molecular Medicine Platform of the University of Medicine and Pharmacy in Iasi.

General criteria HCP – human

Teaching Staff	Center	Qualification
Prof. Puiu Maria	Timisoara	Medical Genetics and Paediatrician - MD, PhD Professor of Genetics, Coordinator of the Genetics Discipline in the University of Medicine Timisoara, President of Romanian Society of Medical Genetics Coordinator of Timis Regional Center of Genetics
Adela Chirita Emandi	Timisoara	Medical Genetics, Paediatrics- MD, PhD Assistant Professor Genetics Discipline in the University of Medicine Timisoara Secretary of Romanian Society of Medical Genetics
Nicoleta Andreescu	Timisoara	Medical Genetics- MD, PhD Assistant Professor Genetics Discipline in the University of Medicine Timisoara
Simona Farcas	Timisoara	Medical Genetics- MD, PhD Lecturer Genetics Discipline in the University of Medicine Timisoara

General criteria HCP – human resources

Teaching Staff	Center	Qualification
Dorica Dan	Zalau	Trainer, Psychologist
Florina Breban	Zalau	Trainer, Legal Advisor
Zsuzsa Almasi	Zalau	Trainer, Psychologist
Mia Acaralitei	Zalau	Trainer, social worker
Dorisz Veres	Zalau	Trainer, social worker
Emese Darko	Zalau	Trainer, Psychopedagogy
Diana Miclea	Zalau	Medical Genetics and Pediatrician - MD

General criteria HCP – human resources

Teaching Staff	Center	Qualification
Cristina Rusu	Iasi	Clinical Geneticist, Pediatrician - MD, PhD Professor of Genetics, Coordinator of the Doctoral School in the University of M Pharmacy Iasi; Coordinator of Orphanet Romania; Coordinator of Iasi Medical Genetics Center, "Sf. Maria" Children's Hospital; Coordinator of the Molecular Lab working with Iasi Medical Genetics Center, "Sf. Maria" Children's Hospital; President of Iasi Subsidiary of the Romanian Society of Medical Genetics;
Lacramioara Ionela Butnariu	Iasi	Clinical Geneticist - MD, PhD Senior lecturer in the University of Medicine and Pharmacy Iasi; treasurer of Iasi Subsidiary of the Romanian Society of Medical Genetics;
Elena Emanuela Braha	Iasi	Clinical Geneticist, Endocrinologist - MD, PhD Senior lecturer in the University of Medicine and Pharmacy Iasi;
Monica Cristina Panzaru	Iasi	Clinical Geneticist - MD, PhD Senior lecturer in the University of Medicine and Pharmacy Iasi; Information scientist for Orphanet Romania;
Roxana Popescu	Iasi	Clinical Geneticist, Molecular Genetics specialization - MD, PhD Lecturer in the University of Medicine and Pharmacy Iasi;

General criteria HCP – human resources

Teaching Staff	Center	Qualification
Eusebiu Vlad Gorduza	Iasi	Clinical Genetics, Cytogenetics specialization - MD, PhD Professor of Genetics, Coordinator of the Medical Genetics Discipline in the University of Medicine and Pharmacy Iasi; Coordinator of the Medical Genetics Unit, "Cuza Voda" Maternity in Iasi; Coordinator of the Cytogenetics Lab working with Iasi Medical Genetics Center, "Sf. Maria" Children's Hospital; Vice President of the Romanian Society of Medical Genetics;
Mihaela Gramescu	Iasi	Biologist, Cytogenetics specialization, PhD Cytogenetics Lab working with Iasi Medical Genetics Center, "Sf. Maria" Children's Hospital;
Oana Pavaloaia	Iasi	Clinical Geneticist - MD; Resident in training (2nd year);
Irina Rezmerita	Iasi	Clinical Geneticist - MD; Resident in training (2nd year);

General criteria HCP – human resources

Teaching Staff	Center	Qualification
Mihai Ioana	Craiova	Medical Genetics - MD, PhD, Coordinator of Dolj Regional Centre of Medical Ger Associate Professor Cell and Molecular Biology; Vice-Dean of Faculty of Medicine
Florin Burada	Craiova	Medical Genetics - MD, PhD Associate Professor Medical Genetics Head of Medical Genetics Department of University of Medicine and Pharmacy of Craiova
Nicolae Cernea	Craiova	Obstetrics and Gynecology - MD, PhD Professor of Obstetrics and Gynecology President of the Obstetrics and Gynecology Advisory Panel of Health Ministry
Stefania Tudorache	Craiova	Obstetrics and Gynecology - MD, PhD Prenatal Genetics; Certificate of competence in Maternal Fetal Medicine; Congenital Anomalies and Fetal Echocardiography
Dominic Iliescu	Craiova	Obstetrics and Gynecology - MD, PhD First trimester genetic and anomaly scan Second trimester anomaly scan; Congenital Anomalies and Fetal Echocardiography
Costin Berceanu	Craiova	Obstetrics and Gynecology - MD, Ph Certificate of competence in Maternal Fetal Medicine; Congenital Anomalies and Fetal Echocardiography
Ioana Streata	Craiova	Medical Genetics - MD; Clinical and molecular genetics, mainly in prenatal diagnosis, pediatrics and neurogenetic disorders associated with intellectual disabilities; Training programme in Clinical Genetics and Neurogenetics

General criteria HCP – human resources

Teaching Staff	Center	Qualification
Marius Bembea	Oradea	Medical Genetics - MD, PhD, Professor of Medical Genetics
Claudia Jurca	Oradea	Medical Genetics - MD, PhD Coordinator of Bihor Regional Centre of Medical Genetics

MINISTERUL SĂNĂTĂŢII
CABINET MINISTRU

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CABINET MINISTRU
Nr. V.V.V. 9382
Ziua 21 Luna 06 Anul 2016

Date: 21.06.2016

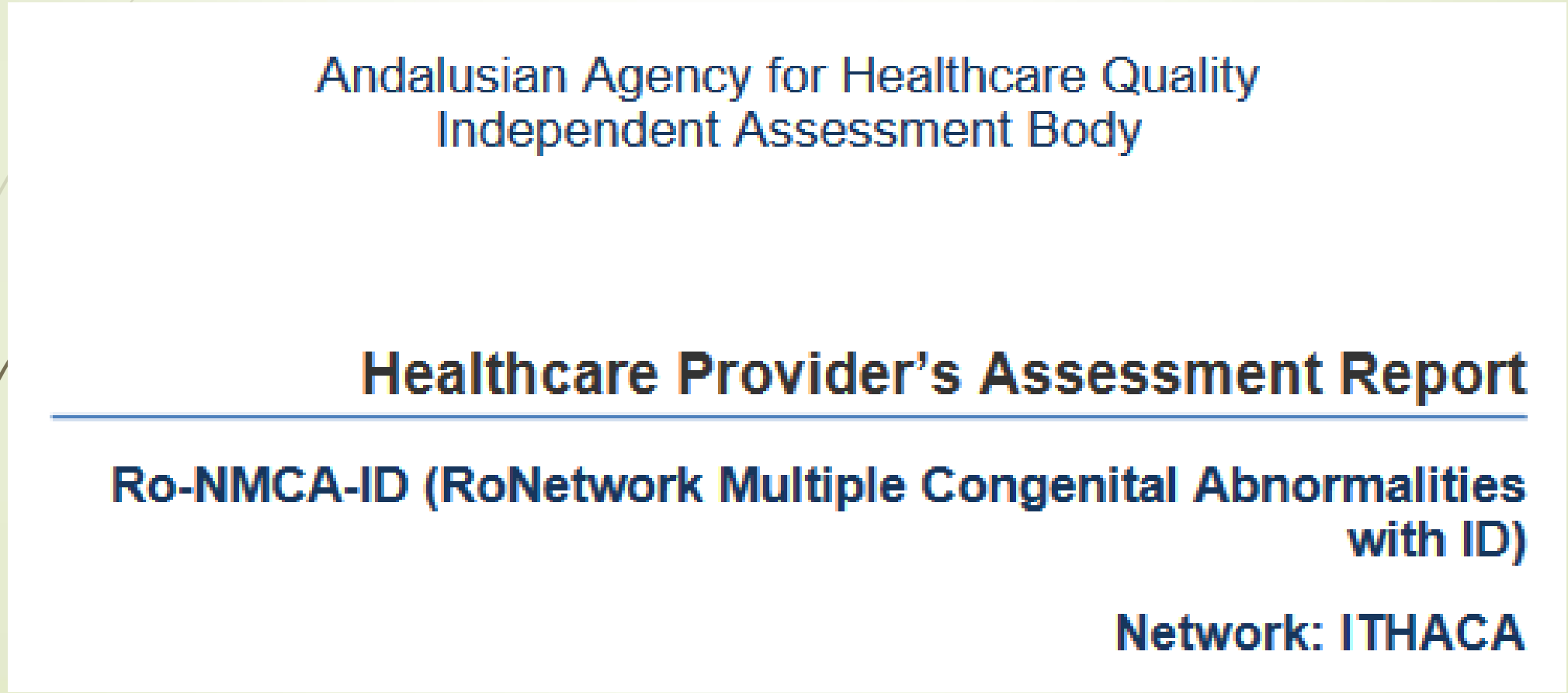
Country: ROMANIA

This letter confirms that the following 5 centers have formed the Romanian Network Ro-NMCA-ID (RoNetwork Multiple Congenital Abnormalities with ID):

1. Regional Centre for Medical Genetics Timis Louis.Turcanu Paediatric Emergency Hospital Timisoara
2. NoRo Centre for Rare Diseases Zalau;
3. Regional Centre for Medical Genetics Dolj- County Emergency Hospital Craiova
4. Medical Genetics Department- "Sfanta Maria" Paediatric Emergency Hospital Iasi,
5. Regional Centre for Medical Genetics Bihor- Municipal Hospital "Dr. Gavril Curteanu"

in order to participate to **European Reference Networks (ERN) for rare diseases,** in accordance with the Romanian legal and regulatory requirements.

ERN-ITHACA = ERN on Rare Congenital Malformations and Rare Intellectual Disability



Andalusian Agency for Healthcare Quality
Independent Assessment Body

Healthcare Provider's Assessment Report

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

Network: ITHACA

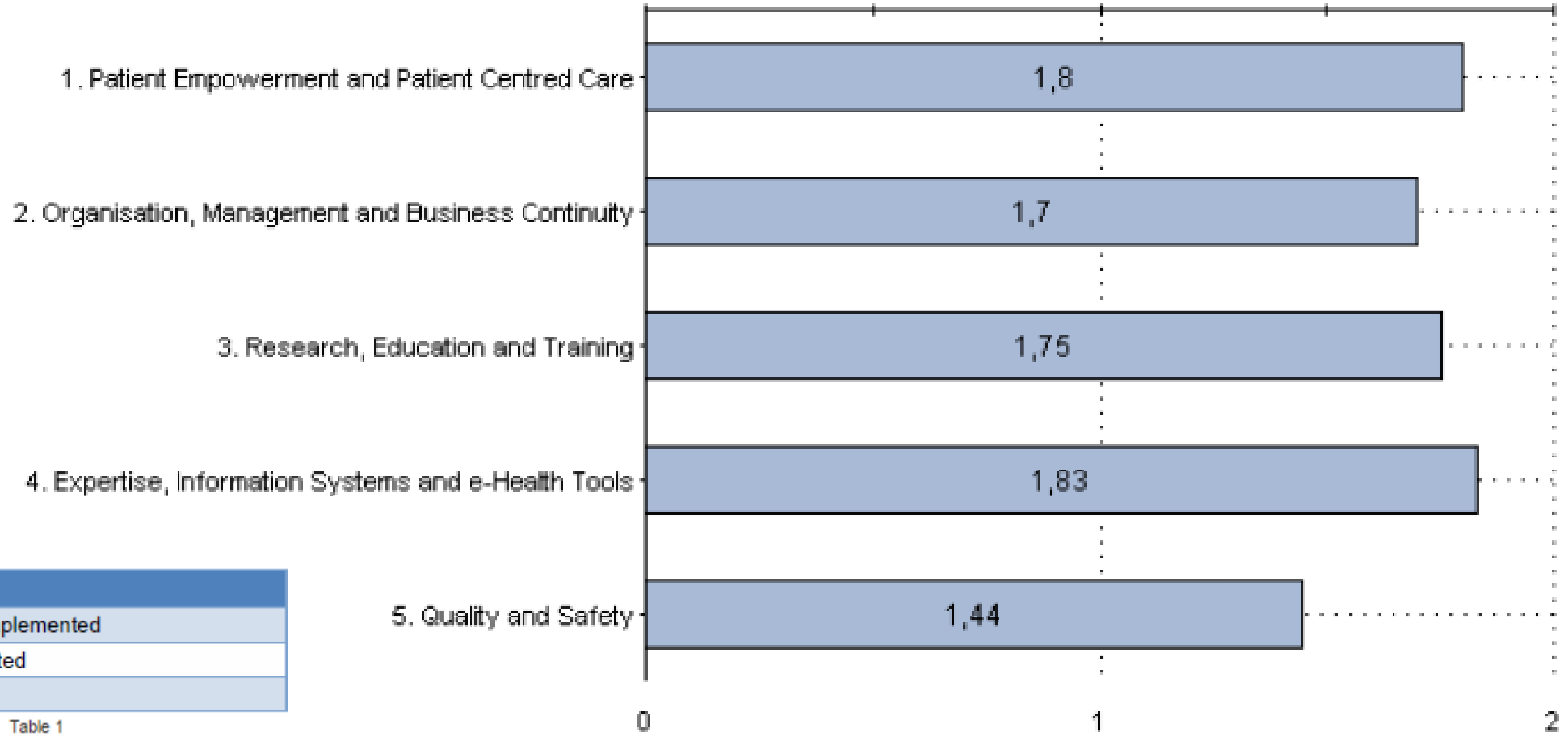
On-site Audit Centrul Regional de Genetica Timis

28.10.2016

- Barbara Kutryba
- National Centre or Quality Assessment in Health Care. (Poland)
- WHO Collaborating Centre for Development of Quality and Safety in Health Systems

- Marc Baaden
- National Center for Scientific Research (CNRS). (France)

Healthcare Providers' Compliance by Theme. General Criteria and Conditions

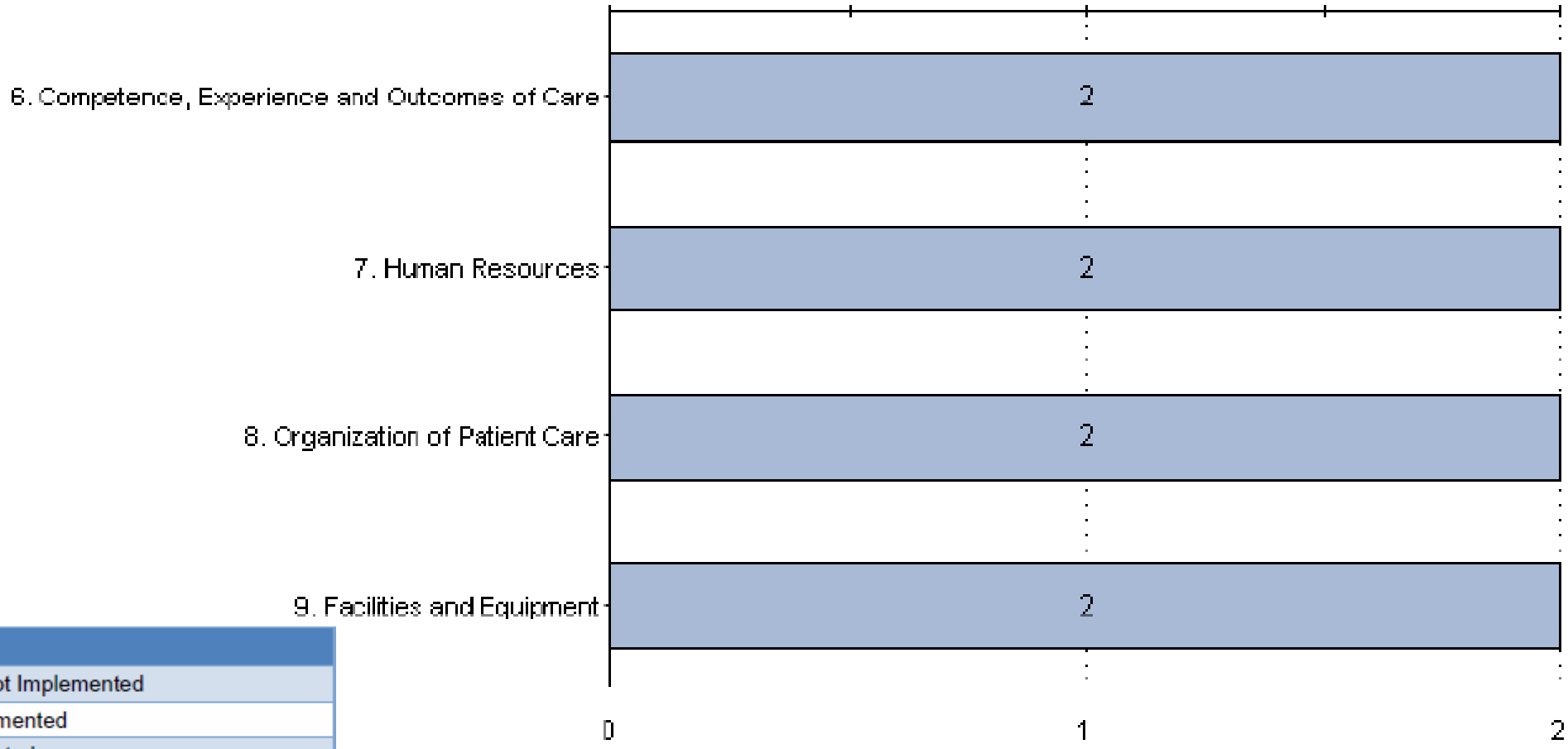


Legend
0: No Activity / Not Implemented
1: Partially Implemented
2: Fully Implemented

Table 1

Chart 2

Healthcare Providers' Compliance by Theme. Specific Criteria and Conditions



Legend
0: No Activity / Not Implemented
1: Partially Implemented
2: Fully Implemented

Table 1

Overall			
Grand Total out of a Possible 140	125	Percent of Total	89.29%


WE DID IT!


now
what?
tomorrow
yesterday

Participating Centres in ITHACA

- 38 centres
- 14 member states
- Coordinator Professor Jill Clayton-Smith

Professor of Medical Genetics, University of Manchester

Clinical Geneticist, St Mary's Hospital, Manchester

Strengths

- Close association with patients
- Existing collective expertise of the centres
- Wide geographical coverage
- Previous experience of some centres with Dyscerne
- Existing networks in some countries
- Existing patient, teaching and training materials
- Strong research track record with some existing collaborations
- We are in an era of major diagnostic developments
- Our plans to share responsibilities

Weaknesses

- Starting from scratch with our network
- Limited funding – EU funds have not come on line as yet
- Need to develop IT
- Multiple languages
- Few validated outcome measures for our type of activities

Opportunities

- Possibility to engage with new centres
- Chance to work in collaboration and not competition
- Broad expertise over our centres
- Existing resources which we can pool and adopt
- Good patient engagement to facilitate development of outcome measures

Threats

- Problems with coordinating such a large network (other centres planning to join in the next wave)
- Limited resources
- IT not yet ready
- Brexit

Objectives/Deliverables For Yr 1

1	Set up and run ERN	Posts recruited. All HCPs active by 6m No of WP with patients involved
2	Establish clear communication channels using IT	6 Teleconferences with minutes Website functional Report on website stats
3	Identify outcome measures	5-10 meaningful outcome measures defined
4	Define disorders where guideline exists and those where needed	Directory of existing guidelines published List of prioritised guidelines for development
6	Identify any existing patient registries	List of existing patient registries and details List of specific registries we have been involved or interacted with
7	Collaborate to add value to research currently being undertaken	Directory of research studies being undertaken within the ERN %age of those where there is collaboration
8.1	Collate and share current education/training resources	List of existing resources No of those who have utilised
8.2	Define unmet training/education needs	List of prioritised needs for parents and professionals

Thank you!