Rare Diseases in pediatric practice

Survey among pediatricians involved in the European Academy of Paediatrics (EAP)

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Introduction

The European Academy of Paediatrics exists to promote the health of children and young people in Europe. The EAP has the ability to bring a wide range of paediatric practitioners across Europe together to improve care, research, and policy affecting the health of children.
The Convention on the Rights of the Child, adopted by the United Nations, is a principle leading document for all the EAP's engagements[1]. This includes a strong commitment to the principles and practices of disability and chronic care management in childhood and support harmonization and implementation.

The EAP, incorporates the section of paediatrics of the European Union of Medical Specialists (UEMS), which aims to harmonize high quality standards of training and health services of medical specialists in Europe. The European Academy of Paediatrics defines different levels of practice: ***Primary Care Paediatrics*** *deals with newborns, infants, children and adolescents in out patient or ambulatory settings like public health clinics, health centers and solo or group practices, under the National Health Service system or privately. It includes all aspects of prevention, diagnosis, management and rehabilitation, extending to the somatic, psychoaffective and social sphere, as well as health promotion and advocacy.* ***Secondary Care Paediatrics*** *is the management of patients with paediatric problems or condition that require an hospital setting for their diagnosis and / or treatment.* ***Tertiary Care*** *is the management of patients with complex and chronic disorders by highly specialised diagnostic and therapeutic techniques, requiring profound knowledge and special skills, and including relevant clinical research.*
The common basic paediatric training[2] includes normal child development and community medicine (complex disability and specific health needs), as well as specialistic paediatric domains and in parentheses the specific signs and diseases such as cardiology, dermatology (pigmented lesions), diseases of orbita and eyes, endocrine disorders, gastro-enterology, genetics and dysmorphology, haematology (sickle cell disease), nephro-urology disorders (haemolytic uraemic syndrome) mental and behavioral disorders (autism spectrum disorders; indication of genetic tests) , metabolic disorder (screening test), neurology and neuromuscular disorders (neuropathies), pulmonology (cystic fibrosis) and rheumatic diseases. The EAP, as paediatric network, connects the primary/ secondary general paediatric care with a wide range of paediatric subspecialists.

Rare diseases often present with common features in the general paediatric practice. Once a rare disease is diagnosed a multidisciplinary approach involving (sub)specialists as well as the primary care providers is essential.
The EAP intends to help reduce the gaps in diagnosis and care as identified by European Rare Diseases Organization (EURORDIS), the 12,000 voices of patients with a rare disease (2009)[3] and supports public health strategies in accordance with the fundamental rights of the children and the rights of the disabled persons.
The EAP has taken notice of the establishment of the Non Governmental Organization (NGO) Committee for Rare Diseases (United Nations, New York) in 2016. The committee has been established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO). This global rare disease platform's aims are:

- To increase visibility of rare diseases at the global level;

- To extend and share knowledge about rare diseases and their unmet needs;
- To connect NGOs interested in rare diseases and their partners within a global platform;
- To promote international, multi-stakeholder collaboration and actions for rare diseases;
- To align rare diseases as a global priority in public health, research and medical and social care policies.

Many rare, often disabling, conditions affect children. Therefore, the aim of this pilot survey is to explore how paediatricians can extend and share their knowledge of rare diseases to improve the well being of children and their families.

Method

An online survey was performed from May 5th- June 26th (year?) among 59 paediatricians, each known through the EAP, since the establishment of EAP's rare disease working group in 2010, by being a member, attending rare disease working group sessions and /or organizing national rare disease conferences inviting the EAP representatives to contribute.
Ten multiple choice questions were sent through Survey Monkey . No reminders were used.
The ten questions address the following issues:

- Practice of the respondents as defined by the EAP (1)
- Children with (features of) rare diseases in paedriatic practice (2 , 3 , 4 , 5)
- Communication between healthcare providers (6)
- Gathering knowledge about rare diseases (7 , 8)
- European Commission strategies on national experts and European networks (9)
- Possible actions to extend and share knowledge about rare diseases (10)

The data were analyzed descriptively.

Rare Disease Survey Questions and Results

Number of responders, response rate, countries: in total, 25 pediatrians responded (response rate: 42%), of which 13 (22%) the first day.

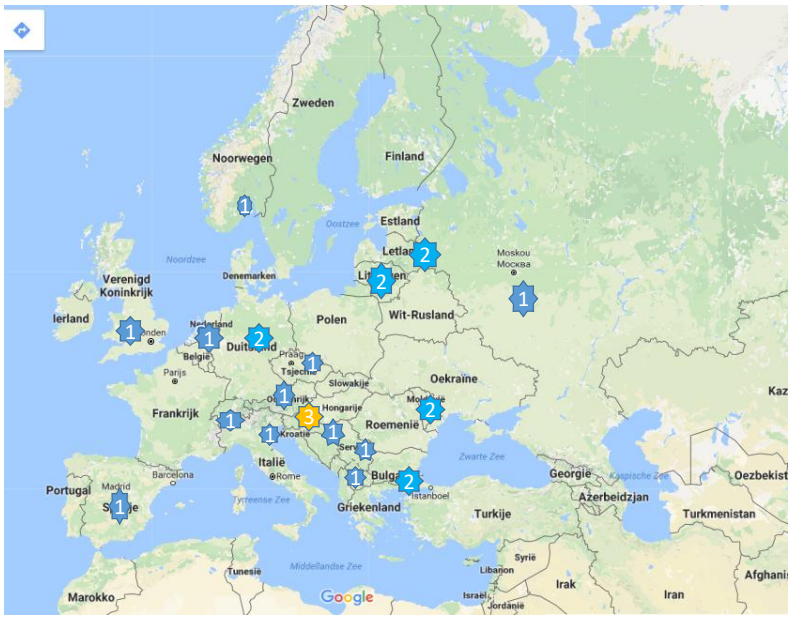


Figure 1. Map with number of responders per countries

Figure 1 shows the number of respondents from 18 different countries, of which 12 in- or affiliated with- the European Union. Average time spent to fill in the questionnaire was 5 minutes.

Practice of the respondents as defined by the EAP

Primary Care Paediatrics deals with newborns, infants, children and adolescents in out patient or ambulatory settings like public health clinics, health centers and solo or group practices, under the National Health Service system or privately. Secondary Care Paediatrics is the management of patients with paediatric problems or condition that require an hospital setting for their diagnosis and / or treatment. Tertiary Care is the management of patients with complex and chronic disorders by highly specialised diagnostic and therapeutic techniques, requiring profound knowledge and special skills.

1. I work in	Response Percent /N=25
Primary care / ambulatory pediatrics	20,0% (5)
Secundary care / general hospital	4,0% (1)
Tertiary care/ academic hospital	72,0% (18)
Other	4,0% (1)

Most respondents work in tertiaty care.

Children with(features of) rare diseases in paedriatic practice:

2. How often do you see a child with a rare disease?	Response Percent /N=25
Daily	28.0% (7)
Weekly or more	36,0%(9)
Monthly or more	12,0%(3)
Once or twice per year	16,0%(4)
Almost never	4,0%(1)

The paediatricians working in tertiary care see at least monthly a child with a rare disease in their practice.

Rare diseases are complex disorders composed of sometimes common features and/or diseases:

3. How often do you see a child with possible signs of a rare disease (growth and / or developmental retardation and / or dysmorhic features)?	Response Percent/ N=25
Daily	16,0% (4)
Weekly or more	36,0% (9)
Monthly or more	20,0%(5)
Once or twice per year	28,0%(7)
Almost never	0,0%(0)

Respondents indicated the following (group of) *features that may part of a rare disease occurring in their paediatric practice:*

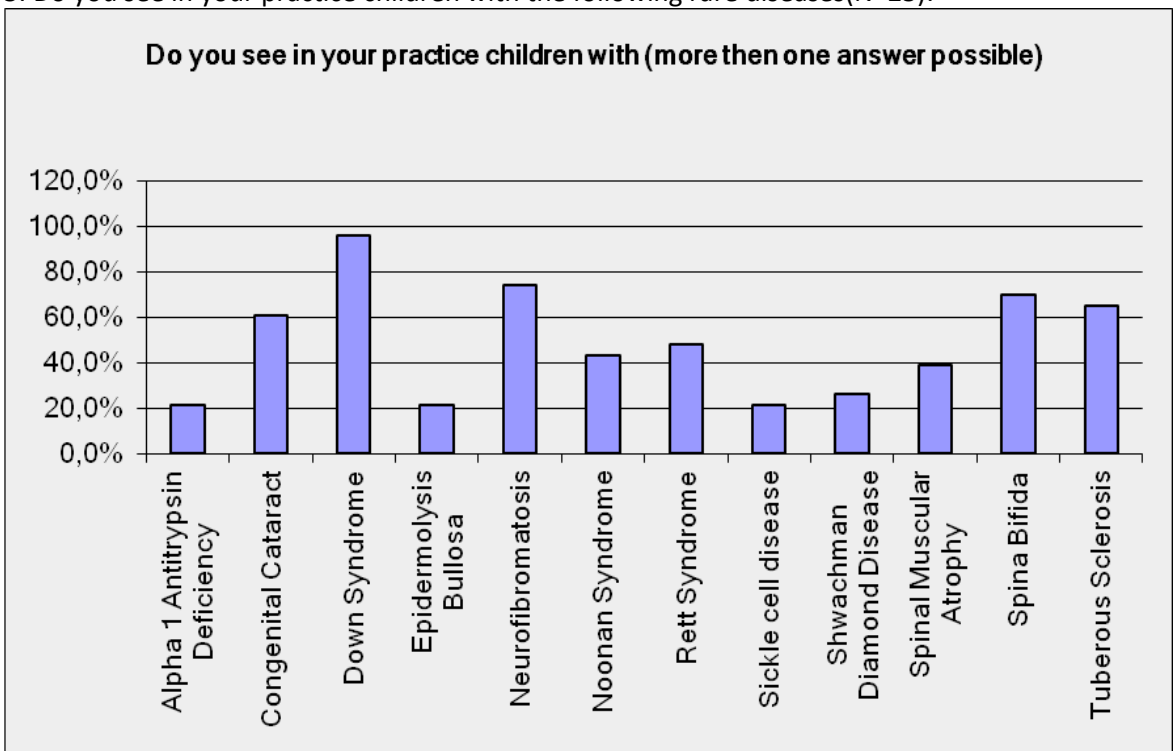
4. Do you see in your practice children with (more than one possible answer)?	Response Percent/ N=25
cancer	44.0% (11)
hematological disease	48,0%(12)
hearing deficit	52,0%(13)
visual impairment	56,0%(14)
kidney disease	56,0%(14)
liver disease	56,0%(14)
endocrine disease	60,0%(15)
pulmonary disease	60,0%(15)
cardiac disease	60,0%(15)

gastro- enterologic disease	60,0%(15)
epilepsy	64,0%(16)
auto inflammatory or auto immune disease	64,0%(16)
skeletal abnormalities	64,0%(16)
autism	68,0%(17)
skin abnormalities	68,0%(17)
neuromuscular disease	72,0%(18)
metabolic disease	84,0%(21)
developmental delay	88,0%(22)
congenital anomalies	88,0%(22)

All of the 6 respondents from primary/secondary care see children with developmental delay and congenital anomalies in their practice.

Twelve different example rare diseases where asked to be known in the respondents practice.
Down syndrome may not be considered as a rare disease, but because of the nature of the syndrome it was included in the survey.

5. Do you see in your practice children with the following rare diseases(N=25):



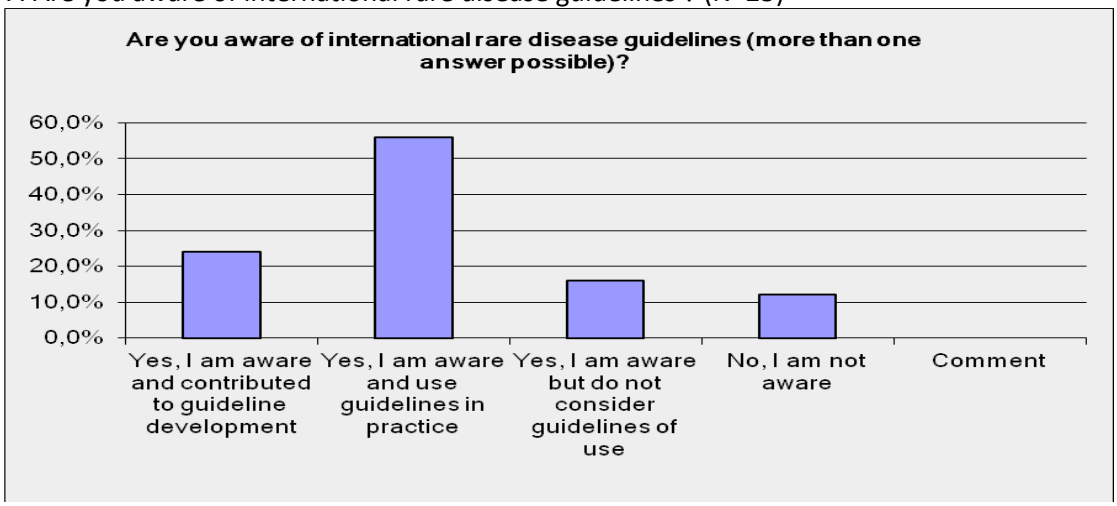
All of these 12 example diseases are represented in one or more of the tertiary care pediatric practices and 10/ 12 diseases in the general practices.

Communication between heathcare providers is essential in diagnosing and caring for a child with a complex disabling condition.

6. How often do you communicate with other heath care providers on the child with a (possible) rare disease?	Response Percent /N=25
Never, I do not see children with a rare disease in my practice	0,0% (0)
Once or twice a year	28,0% (7)
Monthly or more	24,0% (6)
Weekly or more	24,0% (6)
Daily	4,0% (1)
I work in a multidisciplinary team	20,0% (5)

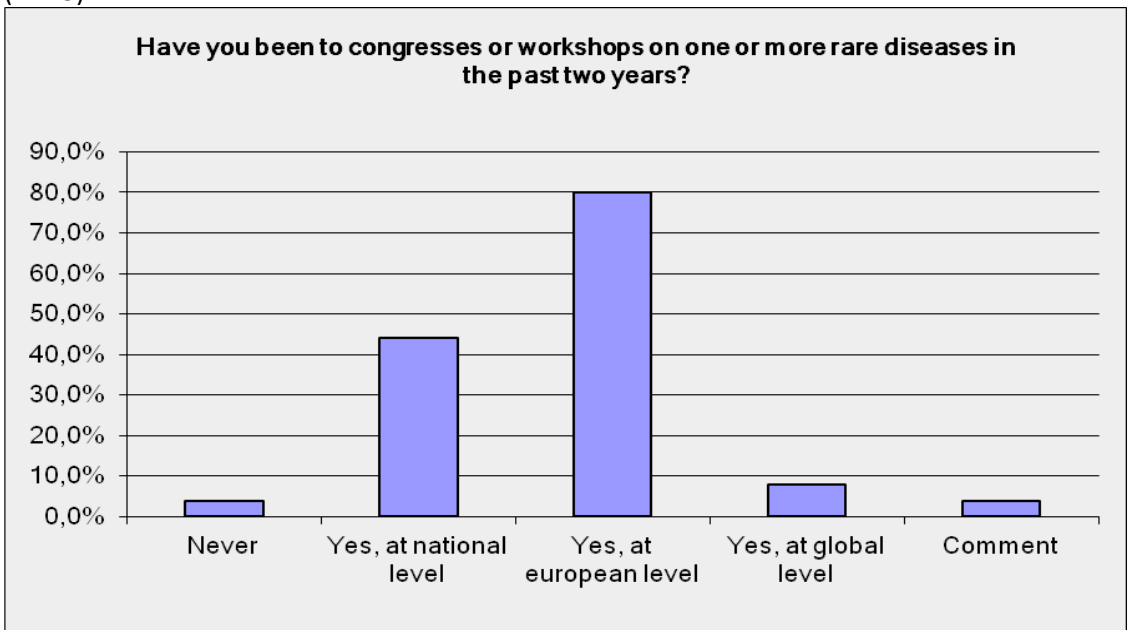
The frequency of the indicated communication is related to the frequency of occurrence in the practice (question 2).

Gathering knowledge about rare diseases
Numerous *rare disease guidelines* are available, for example through rare best practice platform resource[4].
7. Are you aware of international rare disease guidelines ? (N=25)



Most pediatricians are aware of rare disease guidelines and use the guidelines.

Congresses and workshops are organized to enhance knowledge in pediatrics.
Most congresses and workshops are accredited by the national or European specialist societies as Continuing Medical Education (CME) activities established to ensure quality patient care through the harmonization and improvement in the quality of specialists' medical care.
8. Have you been to congresses or workshops on one or more rare disease in the past two years? (N=25)



Most pediatricians attend meetings on one or more rare diseases.

European Commission strategies on national experts and European networks
In March 2017, 24 European Reference Networks (ERNs)[5] were established by the European Commission in frame of the Directive Cross-Border Healthcare. The networks are organized in thematic groups such as haematology, neurology, epilepsy, skin disorders etc. To become part of an ERN the member states have to assigned national experts.

9. The EU member states have assigned national experts on rare diseases. The board of EU member states has approved 24 European Reference Networks (March 2017). Are you aware of the European Union's strategy on rare diseases?	Response Percent /N=25
No, I do not know the national strategy	8,0% (2)
No, I do not know about European Reference Networks	16,0% (4)
Yes, I know how to find national experts on rare disease	20,0%(5)
Yes, I know about the European Reference Networks	56,0%(14)
Yes, I am assigned as a national expert on rare disease	20,0%(5)
Yes, I am particiating in a European Reference Network	16,0%(4)

Over 50% of the respondents, even from non-EU countries, know about European Reference Networks and 20% knows how to find the national experts. Five of these responses came from the 5 non-EU countries. Two general paediatricians in EU countries did not know about their national experts nor of the existence of European Reference Networks.

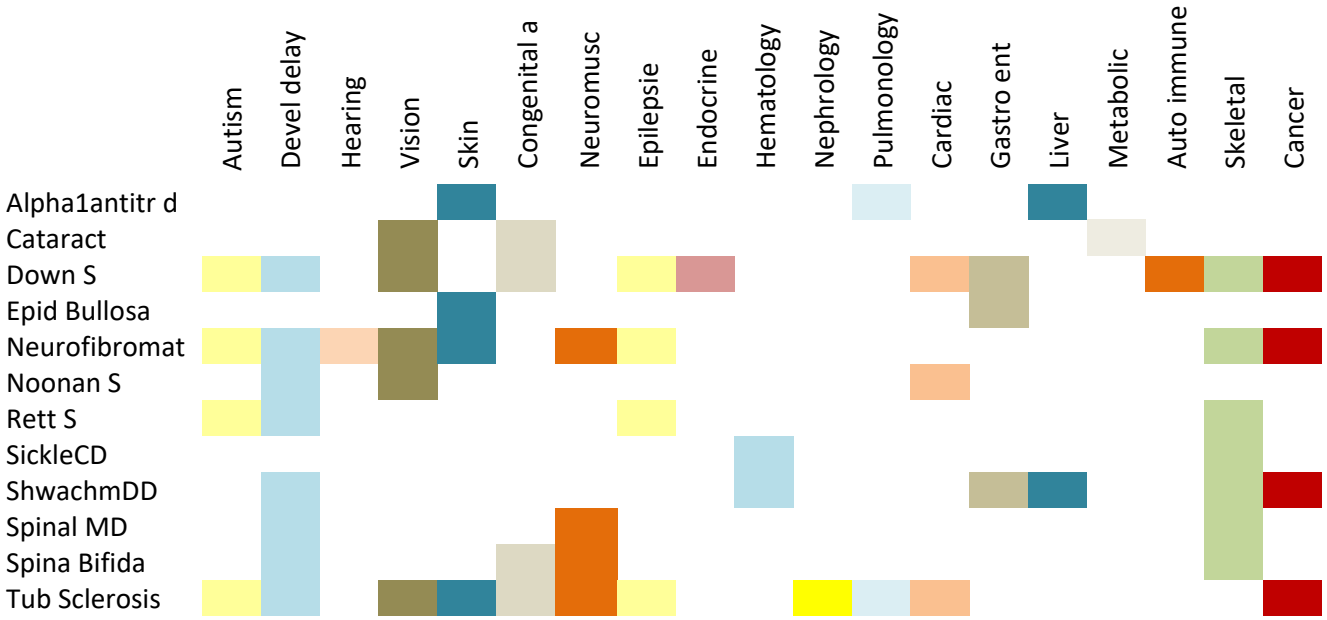
Possible actions to extend and share knowledge about rare diseases
Suggestions of a paediatric response to the **global aim** of Non Governmental Organization (NGO) Committee for Rare Diseases (United Nations, New York) in 2016:

10. What steps are necessary to extend and share knowledge in the field of rare diseases? In my daily practice the care for children with a rare disabling condition (more than one option possible)	Response Percent /N=25
Is sufficient	8,0% (2)
The availlability of more information on rare diseases would enhance diagnosis	72,0% (18)
The availability of diagnostic tests would enhance diagnosis	52,0% (13)
The availability of (international) guidelines would improve care	60,0% (15)
A multidisciplinairy approach would improve care	88,0% (22)
Membership of a European Reference Network would improve care	48,0% (12)

Splitting the responses from general (primary/ secondary) versus sub - specialistic (tertiary) care suggestions differ in the first priority:

Primary/ Secondary	More information on rare and disabling conditions would enhance diagnosis
Tertiary	A multidisciplinary approach would improve care

Rare diseases common in paediatrics
To illustrate the relation between 19 (groups of) features and mono-disciplinary manifestations of rare disease as listed in question 4 is displayed as possible occurring in the 12 selected diseases in question 5 in the table below.



The horizontal line 12 example diseases (question 5) and vertical column the 19 features /diseases (question 4). The colored boxes indicate some of the features/ diseases associated with the rare diseases.
The table illustrates diverse expertise's needed in the care of the child with a rare disease.

Conclusions

Through paediatric training and daily practices paediatricians have experience with features and diseases as part of rare diseases as well as the care for children with a specific rare conditions. Most paediatricians know about (international) guidelines and visit international congresses.
Providing more information at primary care level and the facilitation of co-management between primary secondary care and (sub) specialist (and families) seem to be the next steps to be taken.
It has to be considered the survey was performed among just a small and selected group of paediatricians all known through EAP's Rare Disease network.

Towards comprehensive health care and management for children with rare conditions in Europe[6]:
The World Health Organization states that a collaborative management approach at the primary health care level involving patients, their families and other health care providers may reduce the burden of disease for the child, family, and society. The family-centered chronic care management originated in pediatric care. The EU commission Expert Group on Rare Diseases recommends a multidisciplinary, holistic, continuous, person-centered and participative care to people with rare diseases and full realization of their fundamental human rights. The EAP advocates for international IT standards in open vendor independent IT networks to facilitate person-centered , participative care.

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