Towards integrated paediatric care for rare diseases in Luxembourg: a multidisciplinary and patient-centred study protocol to develop a rare disease diagnostic pathway

Speakers:

Rita Da Rocha Oliveira - CRA & PhD student, CIEC, LIH

Dr. Marianne Becker - Pédiatre, Diabétologue/Endocrinologue, Centre Hospitalier de Luxembourg

Gwennaëlle Crohin - Directrice adjointe, Chef du service de consultation psycho-sociale ALAN

AGENDA

- Rare Diseases and Clinical insights on the barriers around the diagnosis of rare diseases
- Patient Perspective: Challenges and Patient unmet needs during the diagnostic journey
- A multidisciplinary and patient-centred approach to develop a rare disease diagnostic pathway

Rare Diseases and Clinical insights on the barriers around the diagnosis of rare diseases

Over **300 MILLION** people worldwide living with a Rare Disease.

~ 5,000–8,000 unique RDs

~ 80% affect children

~ 30 000 persons in LU

Incidence of Rare Diseases

(Nguengang Wakap S, et. al. (2020) Eur J Hum Genet.; Chung CCY, (2022) Front Public Health); Faye, F, et al. (2024) Eur J Hum Genet;

Case report:

History:

- Uneventful pregnancy, forceps birth at term, birth weight 3080g, birth length 51 cm
- Precocious obesity at the age of 6 months, follow-up at the obesity consultation for 1 year at the age of 3 years
- Normal psychomotor development
- Growth retardation after the age of 4 years
- Headache since the age of 4,5 years

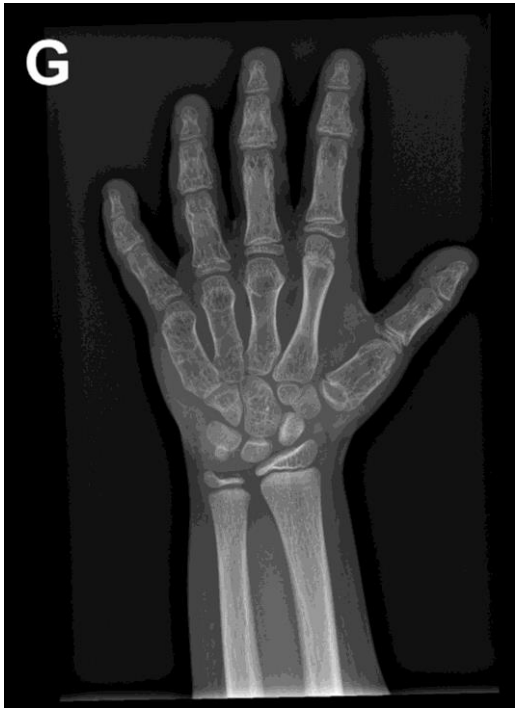
Current history:

- Presentation at the age of 6,5 years in the pediatric endocrine department, as in a blood test prescribed for a cough a low calcium level has been observed

Case report:

Findings:

- *Physical exam:* short fingers and toes, round face, no signs for rachitism
- *Blood test:* **Ca 1.5 mmol/l** (2.2-2.7), Ph 2.93 mmol/l (1-1.8). ALP 280 U/l (142-335), **PTH 375.3 ng/l** (15-65), 25OHD 17.2 ng/ml (30-100)
- X-Ray of the left hand:

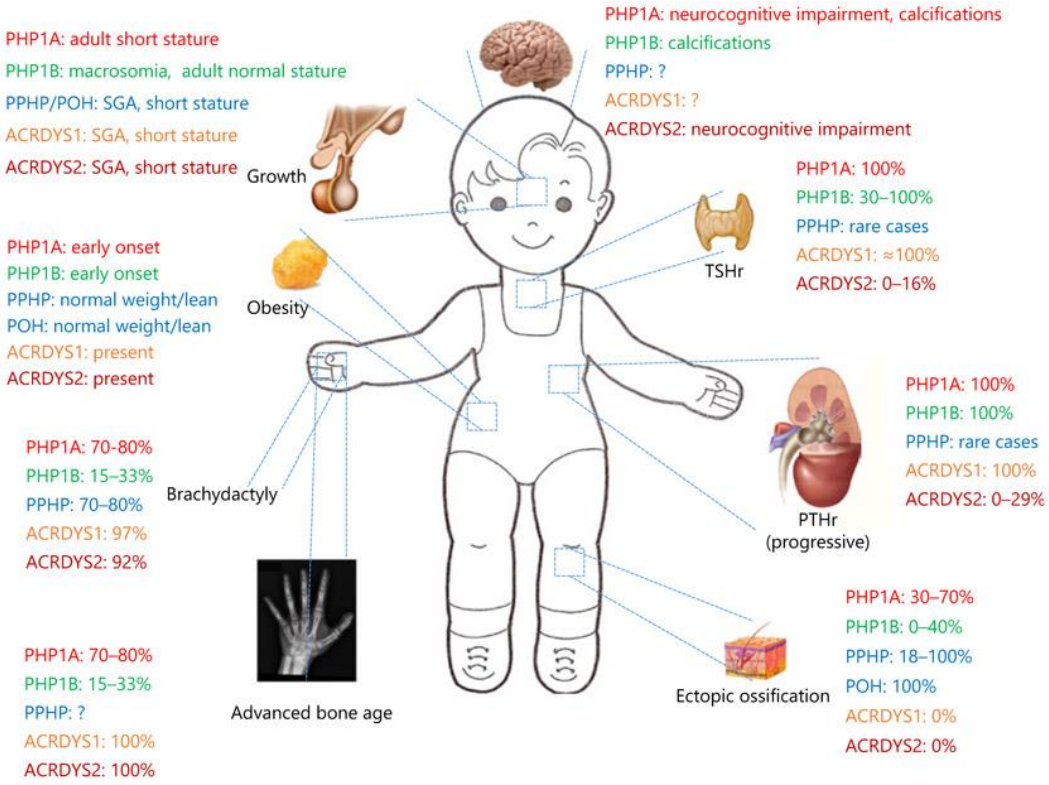


-> Clinical suspicion of a pseudohypoparathyroidism type 1a, confirmed by a genetic analysis

Rare Diseases and Clinical insights on the barriers around the diagnosis of rare diseases

Case report:

Pseudohypoparathyroidism type 1a:



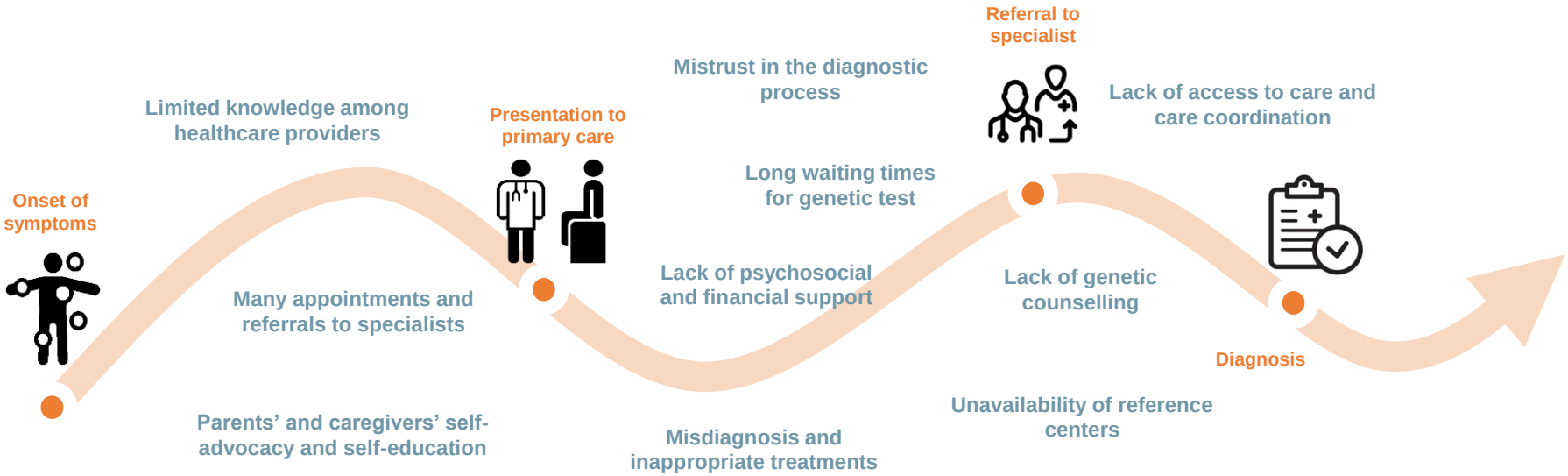
Mantovani G et al. Horm Res Paediatr. 2020;93(3):182-196.

Case report:

Further follow-up:

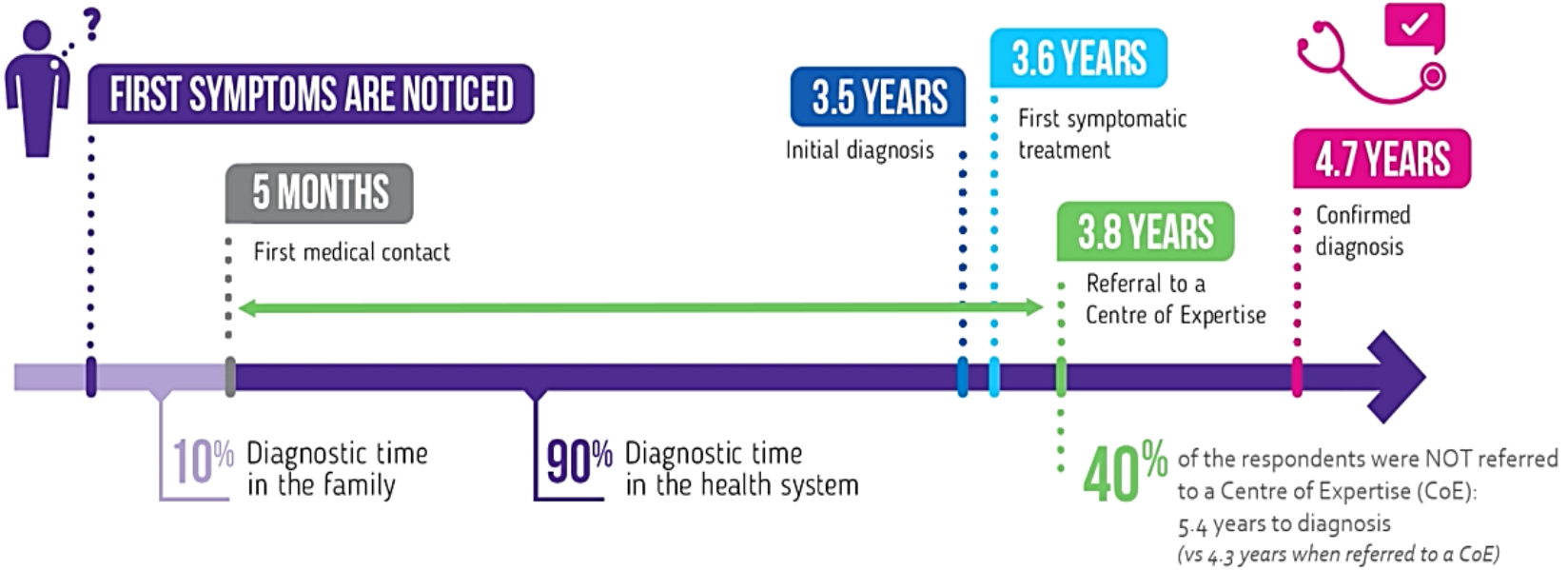
- Treatment with active vitamine D and Calcium -> normocalcemia and dissapearance of headaches
- Screening and treatment of hypothyroidism
- Understanding of the obesity challenge and adpating the caloric input
- Learning difficulties in primary school -> neurocognitive testing: IQ 79 -> organisation of intense support in school and ergotherapy
- Preparation of the family for the reduced adult hight (estimation ca. 140 cm)

Patient Perspective: Challenges and Patient unmet needs during the diagnostic journey



Pavisich K, et. al. (2024) Rare

Patient Perspective: Challenges and Patient unmet needs during the diagnostic journey

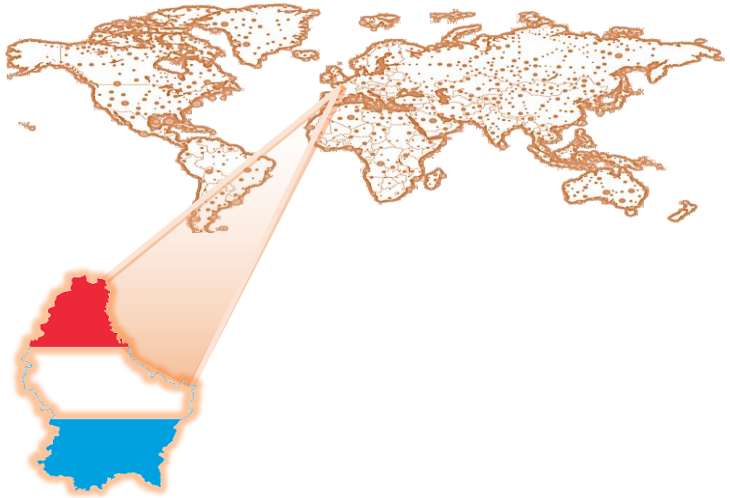


Faye, F., et al. (2024) Eur J Hum Genet

Patient Perspective: Challenges and Patient unmet needs during the diagnostic journey

76 respondents from Luxembourg

- 60% have **consulted more than 5 healthcare professionals** to receive a diagnosis
- **47%** declared lacking psychological support
- **64%** had insufficient coordination of their diagnostic pathway



Faye, F., et al. (2024) Eur J Hum Genet,

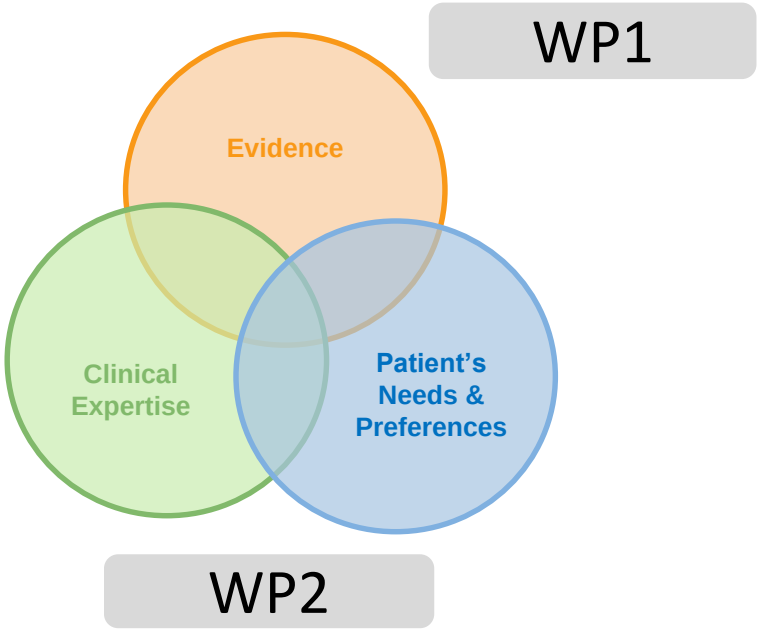
A multidisciplinary and patient-centred approach to develop a rare disease diagnostic pathway

To **develop an integrated diagnostic pathway for pediatric patients with suspected diagnosis of a rare disease**;

To **evaluate the diagnostic experience and unmet needs** of pediatric patients and caregivers;

To **assess healthcare professionals' perspectives (barriers and facilitators** faced during the diagnostic process);

Methodology



Adapted from RarERN Path - Talarico et al. (2020) *Orphanet J Rare Dis*

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Review Questions

Evidence

WP1

- Literature status?
- What are the similarities and differences?
- Does it enhance the quality of care?
- Are these transferable to the Luxembourg healthcare system?

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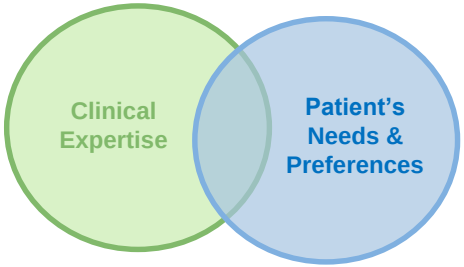
WP2



Qualitative research study

Title: Developing an Integrated Diagnostic Care Pathway Model in Luxembourg for paediatric patients with suspected diagnosis of a rare disease. (L-ICP4Rare-DX)

Sponsor	**Luxembourg Institute of Health (LIH)** **1A-B, rue Thomas Edison** **1445 STRASSEN**
Principal Investigator	Manon Gantenbein, PhD Head of CIEC & CPMO, LIH
Scientific Investigator	Prof. Dr. Jochen Klucken Head of Digital Medicine group (UL)

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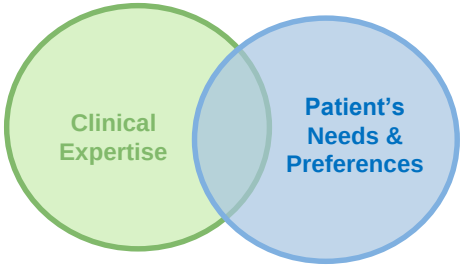
Eligibility Criteria & Topic Guides

WP2

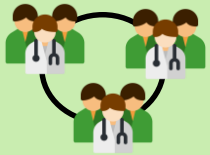


Qualitative research study

Semi-structured Interviews

Patients and their Caregivers/Parents

- Patients and Caregivers **diagnostic journey experience** (including questions about the impact of the diagnostic odyssey in their life);
- Collect **feedback about the unmet needs and preferences** on how the diagnostic journey could be improved;



Focus-Groups

Healthcare Professionals

- **Barriers and facilitators around the diagnostic odyssey** from a healthcare professional perspective;
- **Components that must be integrated to improve the diagnostic process;**

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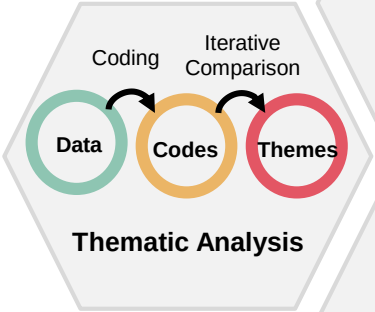
Data analysis



Workshops

Advisory Committee

- Paediatrician;
- Geneticist;
- RD researcher;
- Patient association members;
- 2 patient experts;

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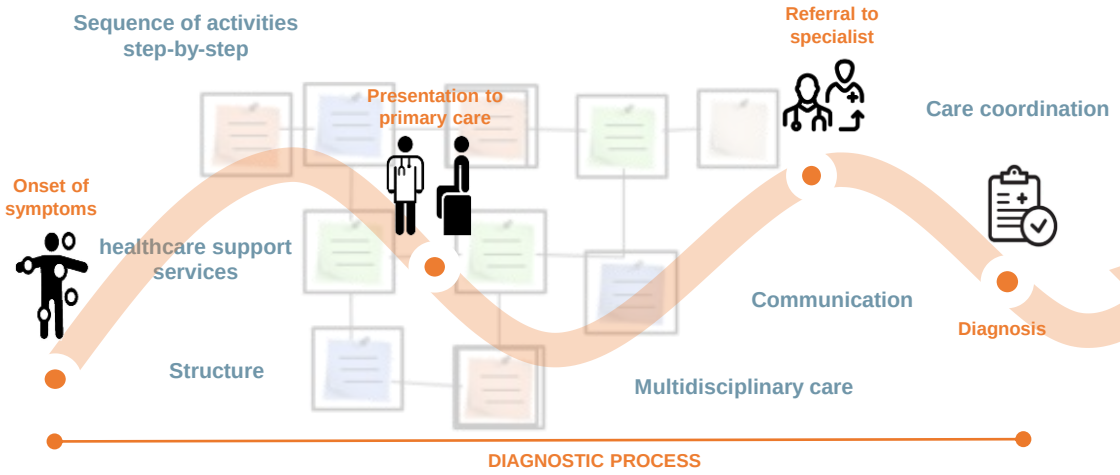
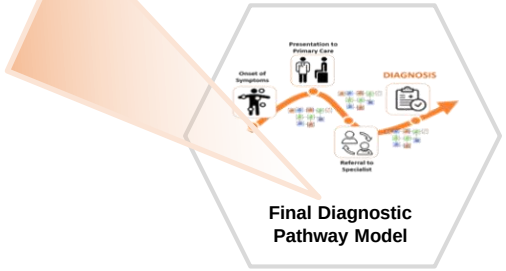
Pediatric patients with suspected diagnosis of a rare disease





EPA defines care pathways aims to:

"To enhance the quality of care by improving patient outcomes, promoting patient safety, increasing patient satisfaction, or optimizing the use of resources"

Thank You!

Rita DA ROCHA OLIVEIRA

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UNIVERSITÉ DU LUXEMBOURG

LCSB

CHL Centre Hospitalier de Luxembourg

LNS LUXEMBOURG

ALAN MALADIES RARES LUXEMBOURG

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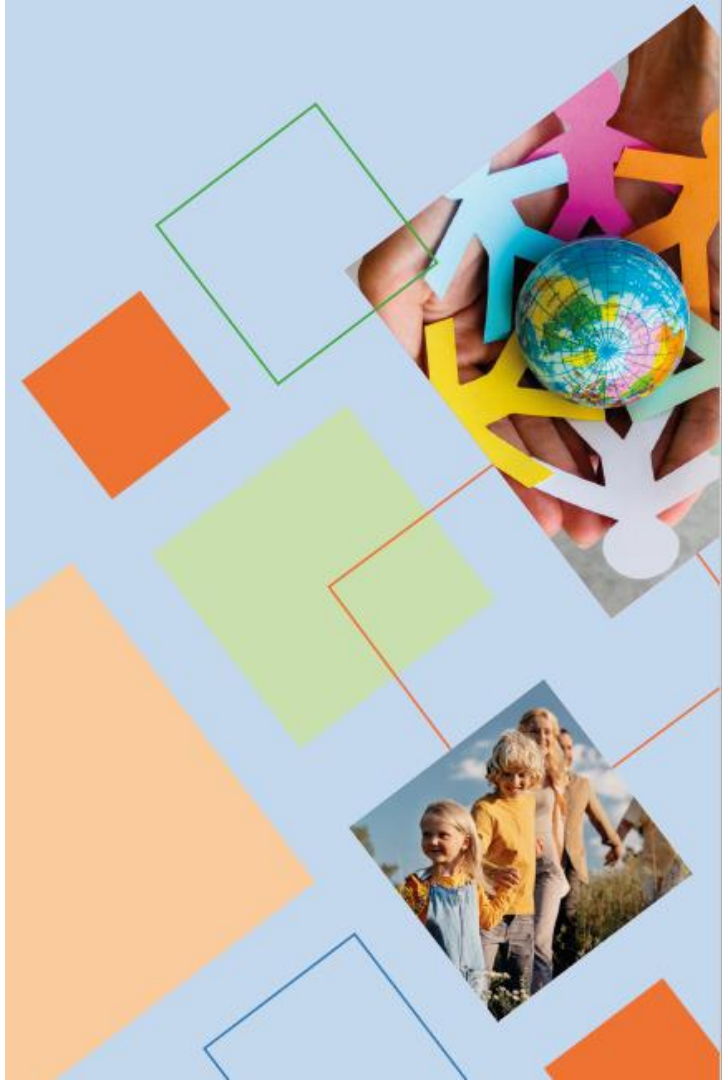

Questions?

Have a look at the **Poster N° 7**

Contact Details

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