



**NF Kinder**

Neurofibromatose durch Forschung besiegen

# Co-Production and Community Participation in Response to Crises

Claas Röhl, founder and chair NF Kinder



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## Agenda

- Rare Diseases - a systemic crisis
- Choosing to become a patient advocate
- Improving health services for NF patients in Austria



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## **Rare diseases - a systemic crisis**

- A disease is considered rare if it affects less than 1 in 2,000 citizens.
- Over 6,000 rare diseases have been identified to date.
- 80% of rare diseases are genetic.
- Rare diseases are often complex and multi systemic diseases.
- Rare diseases affect about 5% of the worldwide population.
- Average time to diagnosis is 4-5 years.
- Only 5% of rare diseases have approved treatments available.

**450.000 people in Austria - 36 Million people in the EU**



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## Rare diseases - a systemic crisis

- Medical expertise is rare.
- Knowledge is scarce.
- Care offerings are often inadequate.
- Research is limited.

Despite their great overall number, rare disease patients are the orphans of health systems. They often lack adequate care and support.

## Choosing to become a patient advocate

- Daughter diagnosed with NF1 at 7month
- Optic pathway gliomas detected at 2 years
- Systemic chemotherapy for 18 month
- Brain surgery at age 3 due to additional gliomas



## NF1 - the disease with the 1.000 faces



CNS  
tibia  
HD,

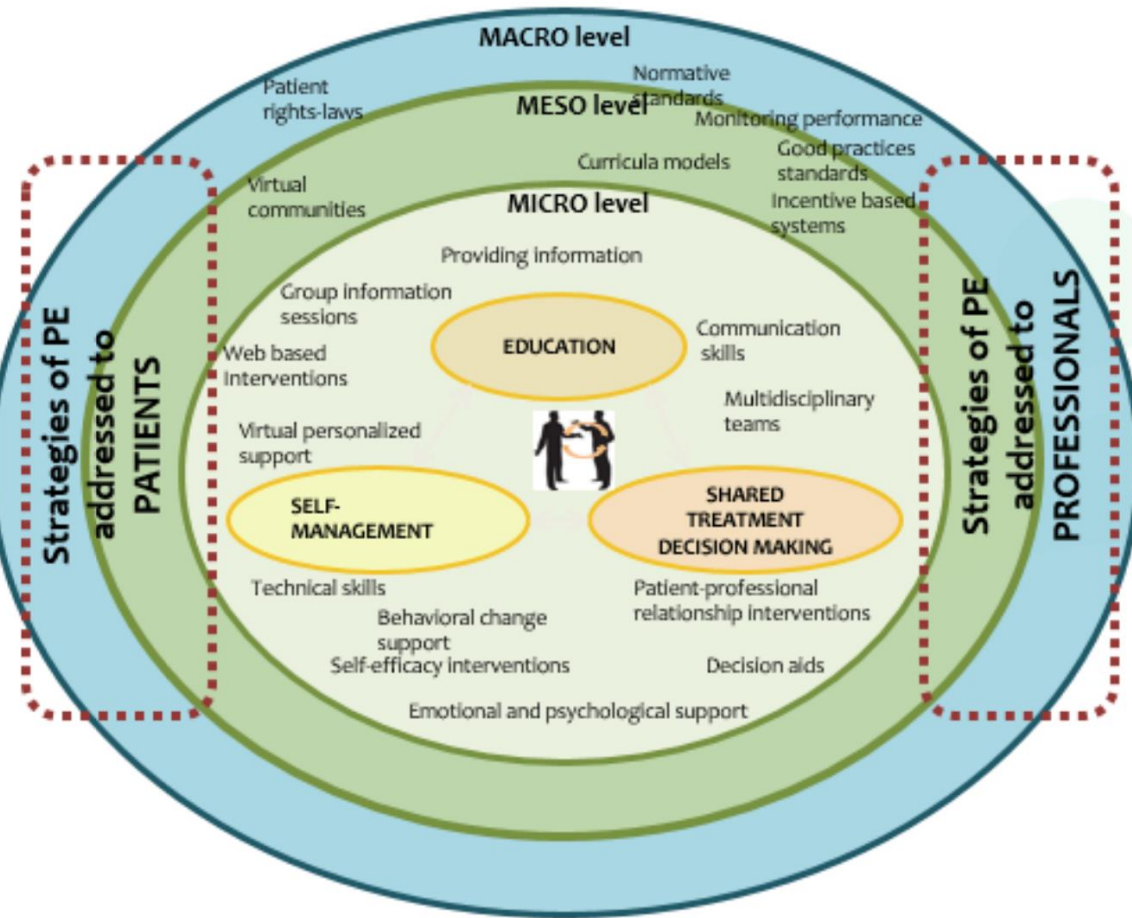


,....)



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- Education
  - Information, health literacy, system factors...
- Self-management
  - Skills, behaviours, self-efficacy, psychological-emotional support...
- Shared decision-making
  - Relationship, decision aids, communication, professional skills, attitudes...



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# Initial situation for NF patients in Austria







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## **Identifying partners - realizing common goals**

Medical University of Vienna - **research, education and patient care**

There is 100% alignment with the goals of our patient organization. By working together we can create a win-win situation.



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## **Patient Care**

NF Kinders vision - a dedicated center of expertise for NF patients

- Diagnosis and counseling
- Routine care (medical and psychosocial)
- State of the art therapies

After entering a contract with the Medical University of Vienna the NF Kinder Center of expertise opened in 2018.



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klin	NEUROFIBROMATOSE Typ 1 (NF1) Pädiatrischer Vorsorgebogen	Intervall:		Zeitpunkte:			
		Alter < 8a	Alter > 8a	Erstvorst.	Kontrolle:	Transition	Kommentar
	Klinische Kontrolle / Anamnese (HW auf ZNS Tu? oder MPNST <sup>1</sup> ?)	mind. 1x/a bzw. bei Verschlechterung		X	X	X	
	Neurostatus	mind. 1x/a bzw. bei Verschlechterung		X	X	X	
	Pubertätsstatus	1x/a + bei Pubertas praecox (♀ <8. / ♂ 9. LJ)		X	X	X	
	Auxiologie (Körperlänge, Gewicht, Kopfumfang)	mind. 1x/a		X	X	X	

# ERN GENTURIS tumour surveillance guidelines for individuals with neurofibromatosis type 1

Charlotte Carton <sup>o</sup> • D. Gareth Evans <sup>q</sup> • Ignacio Blanco <sup>o</sup> • Reinhard E. Friedrich <sup>o</sup> • Rosalie E. Ferner <sup>q</sup> • Said Farschtschi <sup>o</sup> • Hector Salvador <sup>o</sup> • Amedeo A. Azizi <sup>p</sup> • Victor Mautner <sup>o</sup> • Claas Röhl <sup>r</sup> • Sirkku Peltonen <sup>o</sup> • Stavros Stivaros • Eric Legius <sup>o</sup> • Rianne Oostenbrink <sup>o</sup> <sup>o</sup>

On behalf of the [ERN GENTURIS NF1 Tumour Management Guideline Group](#) • [Show less](#) • [Show footnotes](#)

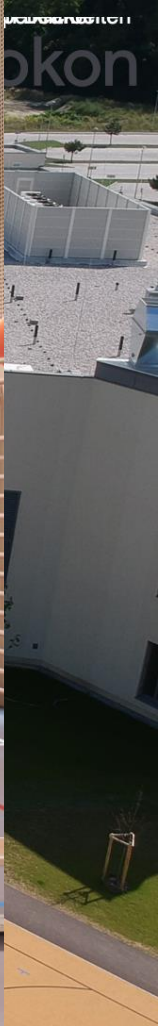
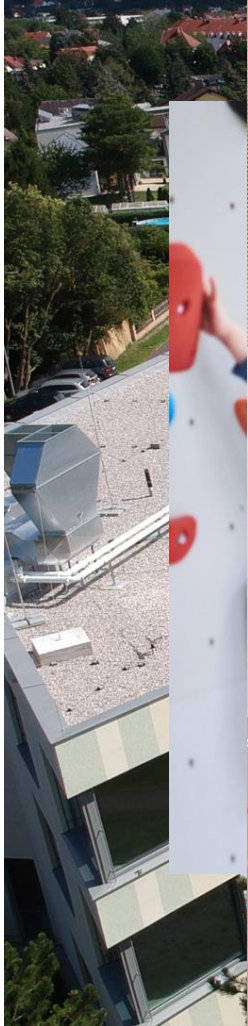
**Open Access** • Published: January 13, 2023 • DOI: <https://doi.org/10.1016/j.eclim.2022.101818> •



### Correspondence information about the author

Ko	Endokrinologie (z.B. bei Pubertas praecox [♀ <8. / ♂ 9. LJ])	bei klin. Notwendigkeit			
	Gynäkologie	1x/a ab Pubertät (Info: Brustkrebs-Screening ab ca. 35 Jahren)			
	EKG/Herzecho	bei klin. Notwendigkeit			
	EEG	bei klin. Notwendigkeit			
	Plastische Chirurgie (z.B. Nerventumoren)	bei klin. Notwendigkeit			
	Kinderchirurgie ( z.B. bei Trichterbrust, Tumoren)	bei klin. Notwendigkeit			

Doppel-Präsentation





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# ← Journal für Klinische Endokrinologie und Stoffwechsel

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## Neurofibromatose Typ 1 – State of the art Betreuung und Unterstützung für Betroffene aller Altersgruppen

verfasst von: Dr. med. univ. Alicia-Christina Baumgartner, Assoc. Prof. Dr. Amedeo A. Azizi

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## **Take home messages:**

Patient organizations should be viewed as an important part in our health care system.

Patient organizations are the gateway to the patient communities and can help other stakeholders to understand the patients perspectives and their needs.

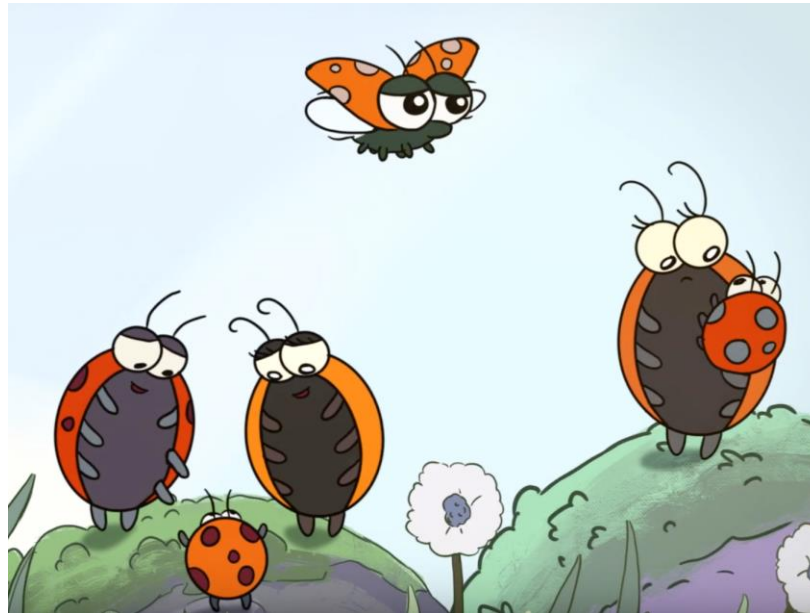
They can help improve health services and make the patient journeys of their community more efficient.

They can help develop clinical guidelines, clinical networks, clinical trial protocols, help recruit patients for studies, organize scientific meetings etc.



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# Danke!