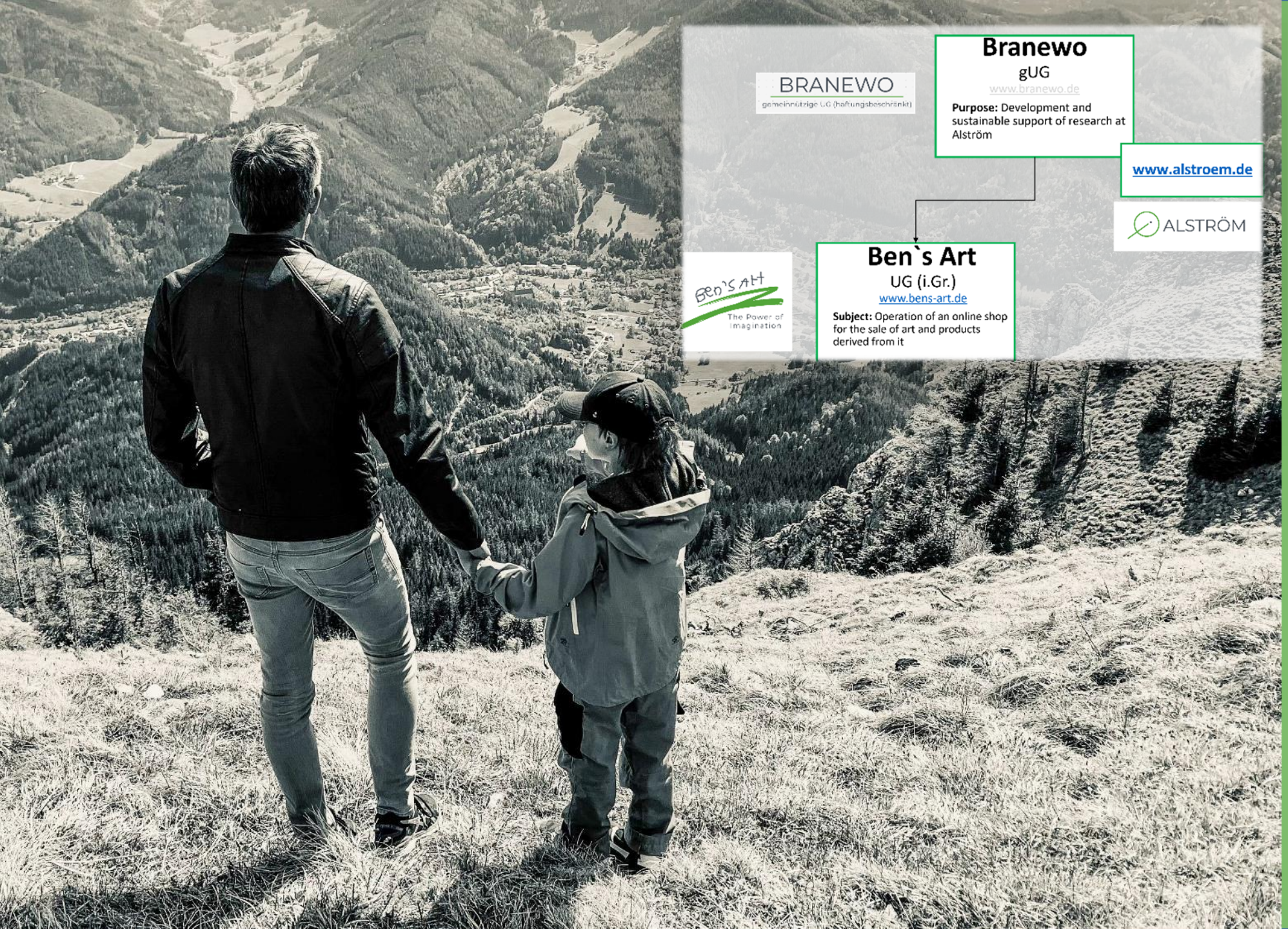


HI,
I am Bernd

DAD & ENTREPRENEUR

Partner with: **PARTNERSEITZ**

TALKING ABOUT THE ALSTROEM SYNDROME

Prevalence: 1:1.000.000 (~1.000 cases worldwide)

Gene Mutation: ALMS1 (‚Large Gene'– e.g., editing not possible)

Knowledge: Little; no pre-diagnosis, difficult diagnosis

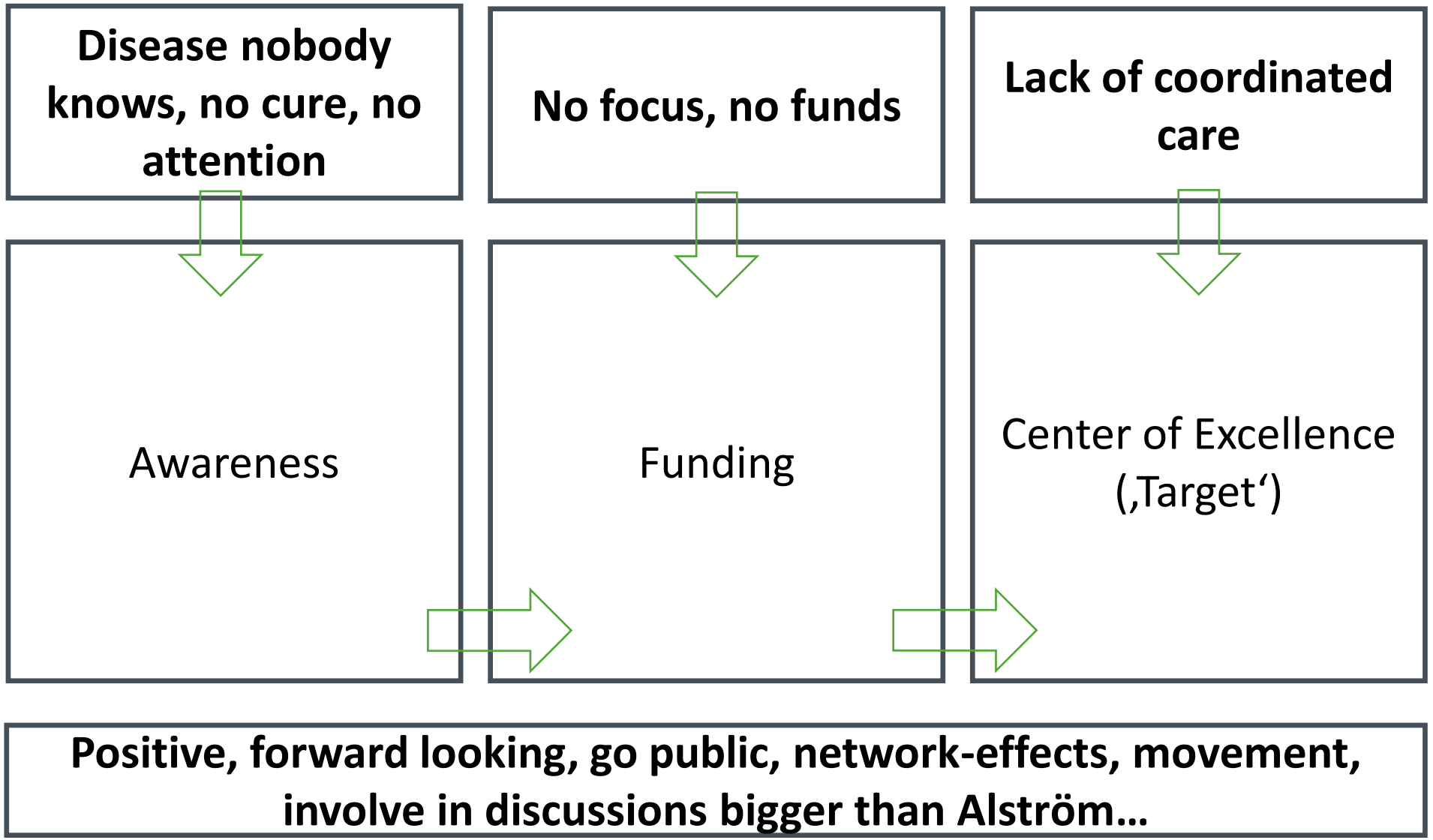
Therapy: Not available, little research

Empirics: Not enough for typical progression of the disease

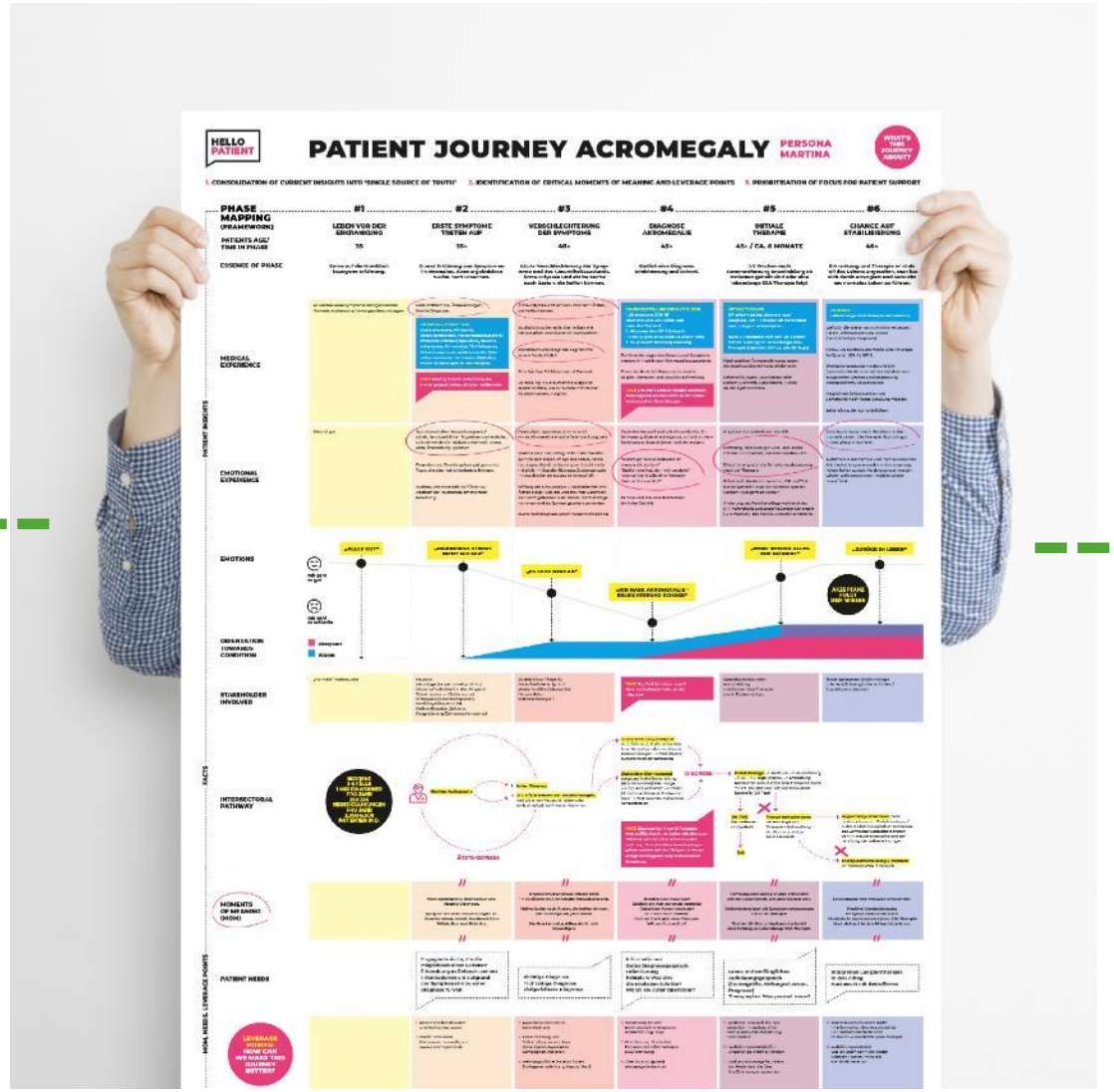
Progression: Especially puberty worsens situation

Category	Seeing 	Hearing 	Organs 	Diabetes 	Other potential diseases <50%
Expression	○ Retinal Degeneration/ Dystrophy ○ Cone dystrophy ○ Nystagmus ○ Photophobie ○ Colorblindness ○ Blindness (over time)	○ Progressive bilateral sensorineural hearing loss	○ Cardiomyopathy ○ Fatty liver ○ Fibriosis ○ COPD	○ No feeling of satiety ○ Obessity ○ Insulin Resistance ○ Hyperinsulinism ○ Diabetes mellitus type 2	○ Respiratory problems ○ Kidney disease, Bladder dysfunction ○ Hyperlipidemia ○ Hypothyroidism ○ Short stature ○ ….
Probability	100% (typically first symptome)	>80 %	>40%	>60%	

MY MISSION? THINK BIG, ACT DIFFERENTLY!



PATIENT JOURNEY IS BASIS FOR ‘THE TARGET’



Center of Excellence (for Alström*)

++ Background ++

Vision:

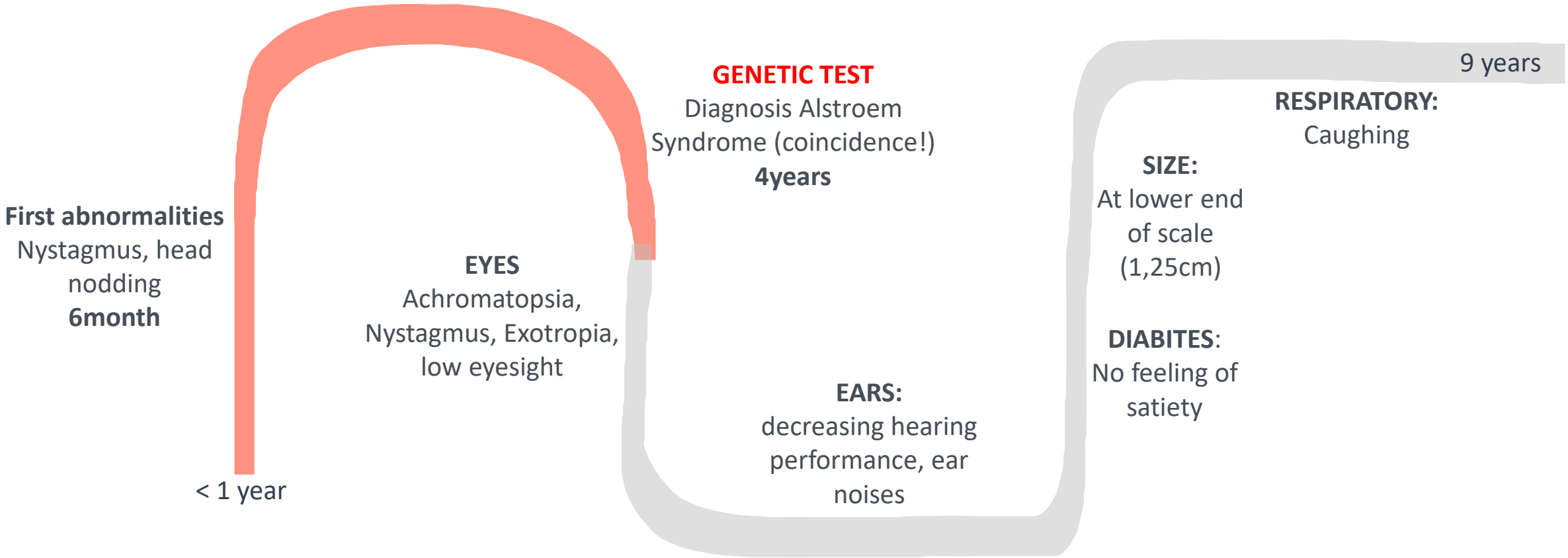
"Navigator" (in the sense of a contact person within a physical facility). This point of contact is immediately delivered by the search engine when searching for the specific disease.

Goal:

The Center of Excellence for (Alström*) is a physical unit ('under one roof'). This ensures an ecological and interdisciplinary, comprehensive support, processes and roles (responsibilities) are clear, necessary changes (DataGovernance etc. are initiated).

(*The starting point and always the first priority in the implementation is the rare disease 'Alström syndrome'. However, the concept should not necessarily be limited to this_)

BENS SYMPTOMES



WHAT ARE THE CHALLENGES AND LEARNINGS?

WHAT'S WRONG?

Looking for answers

Info on symptoms

Searching for a diagnosis

Hopeing for coincidence

Finding the right (midical) contact person

NOW WHAT?

Dealing with a 'rare' diagnosis

Where can I get information about the disease, therapy and support?

Explaining the diagnosis to family, friends and those around me

Studying clinical trials

Communicating with physicians and healthcare providers/reimbursement

IMPACT ON EVERYDAY LIFE

Integrating the disease into everyday life

Caring for a patient with a rare disease

Change of lifestyle

Impact on family, job and leisure

Dealing with the disease

Searching for fellow sufferers/SHG

TRANSITION

Transition from childhood to young adulthood

Independence, relationships & family

Medical transition (change of physician)

FUTURE

Engagement & participation

Sharing one's own experiences

Participating in studies

Hoping for future therapies

Life perspectives

MANAGEMENT & LEARNING

Dealing with misdiagnoses, wrong or no information

Managing disease, therapy and side-effects

Finding the right healthcare partner

Finding appropriate supportive care

Participating in social activities

Acting in critical (medical) situations (e.g. narcosis, do or don't situations)

Decision taking (e.g. growth)

Relevance of social status

Accepting, nobody can tell you if patient is ‚healthy'

Lack of coordinated care

THANK YOU

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