# Patient Journey **Netherton Syndrome** - Ichthyosis Linearis Circumflexa

#### 1. First symptoms Birth/baby

2. Diagnosis Any stage in life

**Risk of dehydration** elevated sodium levels in the blood **Failure to thrive Generalised redness or** trouble gaining weight scaling of the skin Neonatal erythroderma Trichorexis Nodosa, Predisposed to bacterial & Trichorexis invaginata viral infections, both skin & systemic Problems in hair may occur, in different stages. Bamboo hair.

**Predisposition to allergies** (food and *inhαlation*), **asthma and eczema** 

#### Need:

- Period after birth might be traumatic for parents
- Parents face lack of knowledge/information
- Parents should be taught how to take care of the skin
- Parents should learn to recognize symptoms of infections of the failure to thrive
- Parents need a buddy system
- Pyschological support

#### **Ideally:**

• Expertise centers: where are they? How can you contact them

#### Need:

- Information on condition & inheritance & future perspective
- People who can share their own experience
- Psychological support

**3. First treatment** As a baby or when diagnosed later in life

Diagnosis can be made in any stage of live. Hopefully it is soon after birth.

LEKTI absence in skin biopsy

Hairs change

**Topical treatment:** Current treatment options are limited to topical treatment with mild moisturizers containing petrolatum or lanolin and/o a skin barrier repair formulacontaining ceramides or cholesterol.

**Genetic testing** 

**Mutations in SPINK 5 genea**  treatment.

#### **Ideally:**

- Registration
- The testing can rarely tell how mild or severe the condition will be in any particular individual

 Test both for DNA and absence of LEKTI, somebody can have a new mutation leading to absence of LEKTI

 Expertise centers: where are they? How can you contact them?

#### Need:

- There is lack of evidence of treatment
- Patients need improvement of their quality of life
- Patients and parents need information in their own language

**Topical steroids:** Are αlso readily absorbed and should be used in low potency forms for short periods of time.

### 4. Other sympthoms

Ectropion or skin cancer might occur. This can be in any stage in life.

> In case of ectropion, eyelid reconstruction.

> > sometimes needed.

**Topical calcineurin:** inhibitors are helpful for individuals with Netherton but must be used sparingly (risk of high blood levels). This can be a painful

**Topical treatment** 

Systemic treatment: Skin barrier Immune modulators, like IVIG and biologicals

#### **Ideally:**

- Guidelines on treatment Patient guideline on Netherton Information in their native language
- Expertise centers: where are they? How can you contact them?

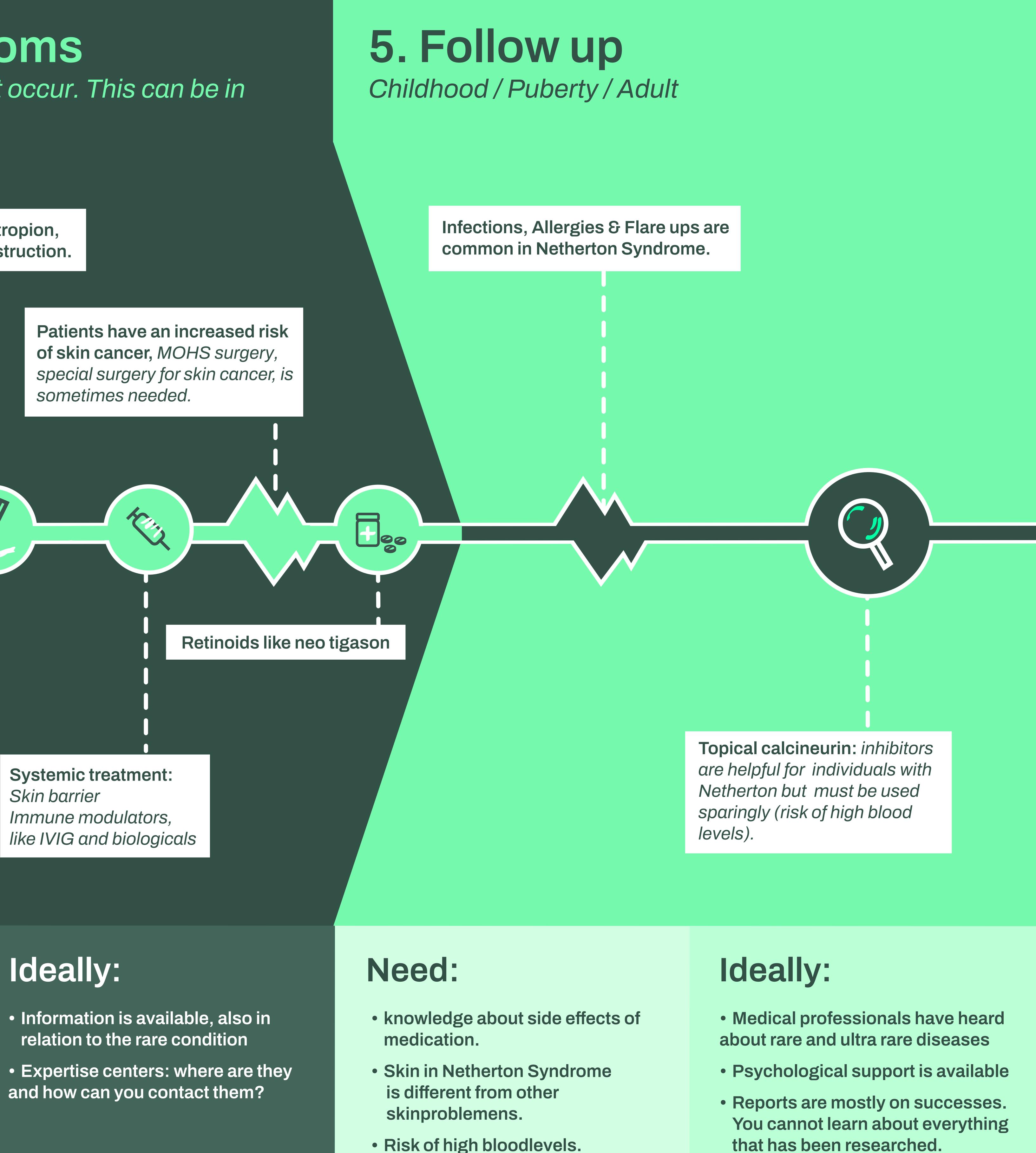
#### Need:

- Information on skincare
- Information on eye care
- Lack of information for care professionals
- Need of multidisciplinary care

#### **Ideally:**

- Information is available, also in relation to the rare condition
- and how can you contact them?

## Care4 Netherton



- - Expertise centers: where are they and how can you contact them?