

Dysmelia and Polands syndrom

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Terms and definition dysmelia

The term "reduction defect" is used for conditions where there is a lack of an arm and/or leg

Dysmelia:

Congenital condition where arms and or legs are not developed as "normal".

dys (Greek) – deficient or "wrong"

melos (Greek) – limb

We will use the term dysmelia instead of reduction defect in the presentation

Dysmelia – many terms in English



congenital

- limb deficiency
- limb anomaly
- limb reduction defect/deformity
- limb amputation

Limb deficiency present at birth

Dysmelia – a rare disorder

In Norway, 4 children were born with dysmelia in 2022

(In the years 2016-2021, 8 – 18 children were born with reduction defects a year (FHI medical birth register))

Most have dysmelia only on the arm, some have dysmelia only on the legs and a few have dysmelia on both arms and legs

TRS has 526 registered users with dysmelia, 140 are 0 - 18 years old (August 2023)

Causes of dysmelia

- We still know little about the cause of dysmelia!
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- Dysmelia occurs early in fetal life (5-11 weeks)
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- 1/3 of the cases are due to disturbances in the blood supply
- 1/3 is due to inheritance or genetic changes
- In 1/3 of the cases, the cause of dysmelia is unknown
- A few cases (approx. 4%) are due to exposure to substances harmful to the fetus in the womb (e.g. Thalidomide)

Eximination of new born babies

/helsebiblioteket.no

Pediatriveiledere

Fra Norsk barnelegeforening.

[Generell veileder](#) > [Revmatologi, ortopedi, skjelett og skalle](#)

4.14 Anbefalinger for utredning av nyfødte med reduksjonsdeformiteter

☰ OVERSIKT

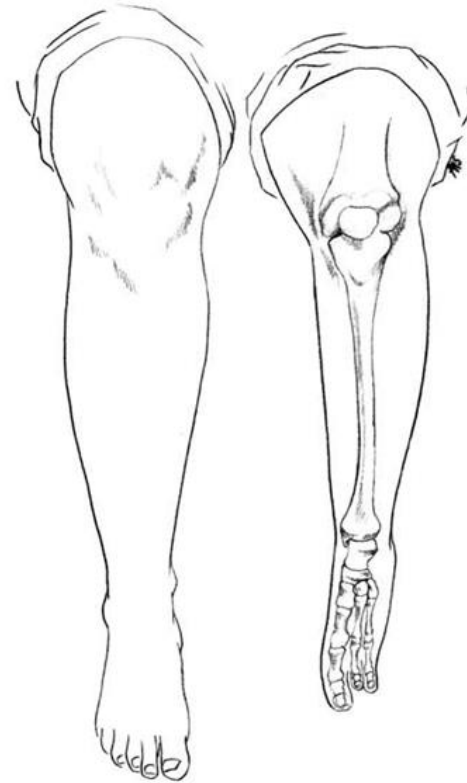
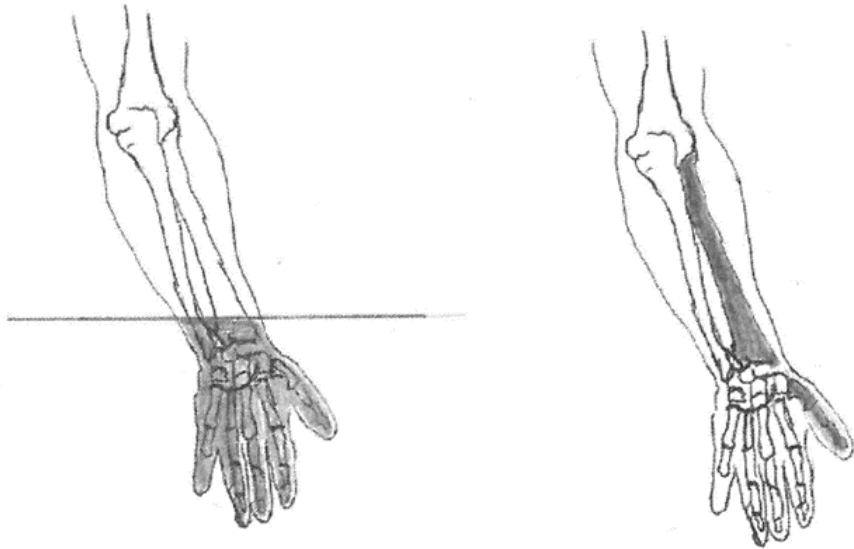
The pediatrician has the primary responsibility for coordinating the investigation of the condition and referring to the dysmelioma team

Pediatric radiologist, orthopedist, hand surgeon and geneticist take part in the diagnostics as needed

- *Film from TRS: [Å få et barn med dysmeli \(youtube.com\)](#)*

Dysmelia - description

- Transverselle (tversgående)
- Longitudinelle (langsgående)

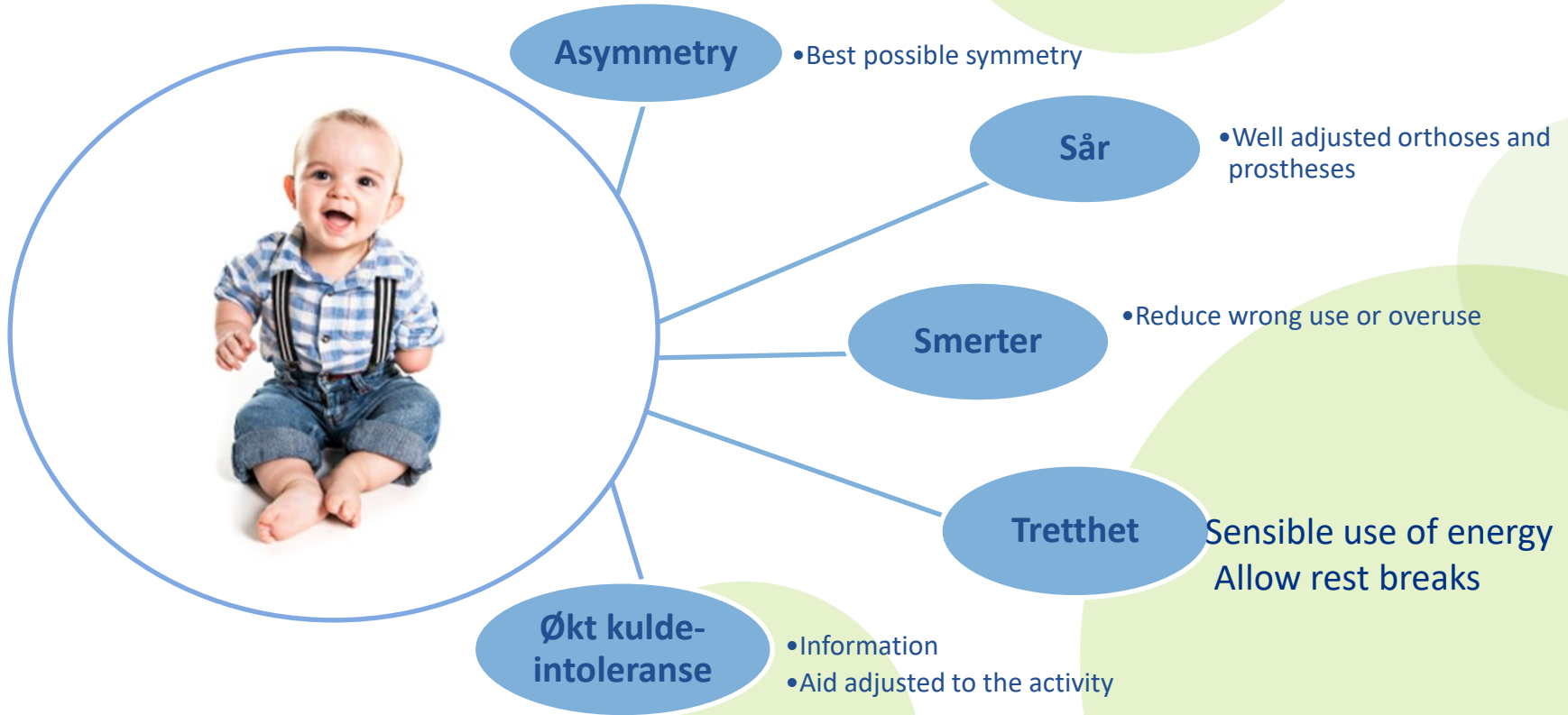


What can be different in the body

- The skeleton; bones in the affected body part are missing, may be smaller or have a different shape than usual
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- Muscles may be missing or less developed
-
- Blood vessels may be less developed
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- The nerves are present, but not always where they usually are



Possible physical challenges



Norske anbefalinger for oppfølging av personer med medfødt mangel på overekstremitet i form av transversal reduksjonsdefekt (en type dysmeli)

<https://app.magicapp.org/app#/guideline/3380>

- In the first year of life, the child and the parents should be offered contact with the dysmelia team at least at 6 months and at 12 months of age
- Children of and under school age should be offered annual interdisciplinary follow-up, to capture changes in needs as they grow and develop.

Dysmelia teams

The pediatrician refers to the dysmelia team

5 dysmelia team for arm/hand dysmelia

Tromsø, Trondheim, Bergen, Oslo and Ottestad/Inlandet

1 dysmelia team at leg dysmelia (Oslo) (National treatment service for congenital deformities in the lower limbs in children)

The Dysmelia teams offer regular checks.

Assesses function, need for prostheses and aids, need for surgical treatment and training

Transfers to local auxiliary device

Prognoses – how does it go?

Life expectancy as long as for the rest of the population

Children with dysmelia cope well - as well as most children

The children do not experience themselves as disabled

Some have to go through operations

Some people get more pain in muscles - bones - joints in adulthood

- Questions?
- Comments?



Ressources

- Fagmiljøer for personer med dysmeli - Sunnaas sykehus HF
- Norsk Dysmeliforening |
- TRS kompetansesenter for sjeldne diagnoser - Sunnaas sykehus HF
- Film for health professionals: Veiledning for dysmelibehandling - YouTube