

Lymphedema in Phelan-McDermid syndrome

Raoul CM Hennekam

Amsterdam UMC, Amsterdam, Netherlands



Topics

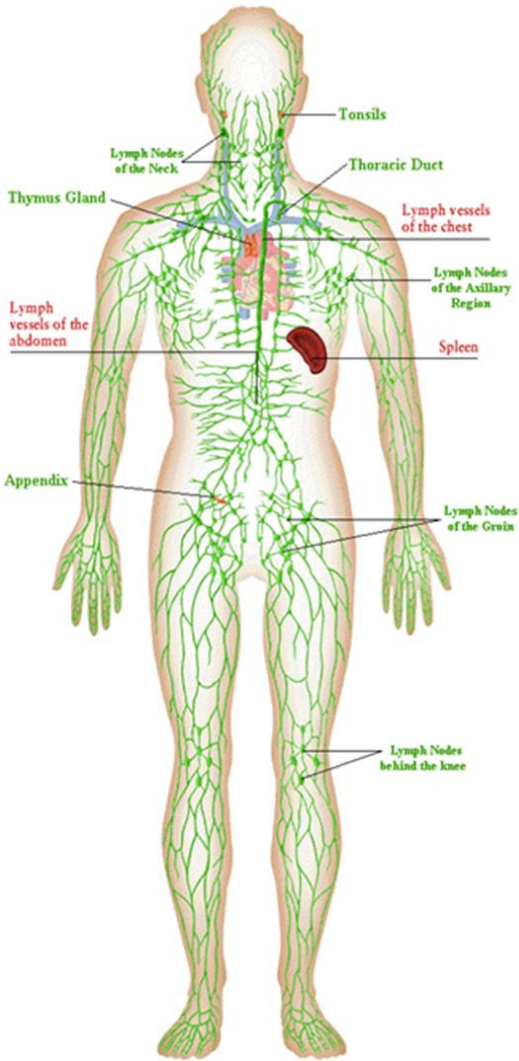
- lymphatic system
- lymphedema
- subtypes
- why in PMS
- age
- site
- static / progressive
- management



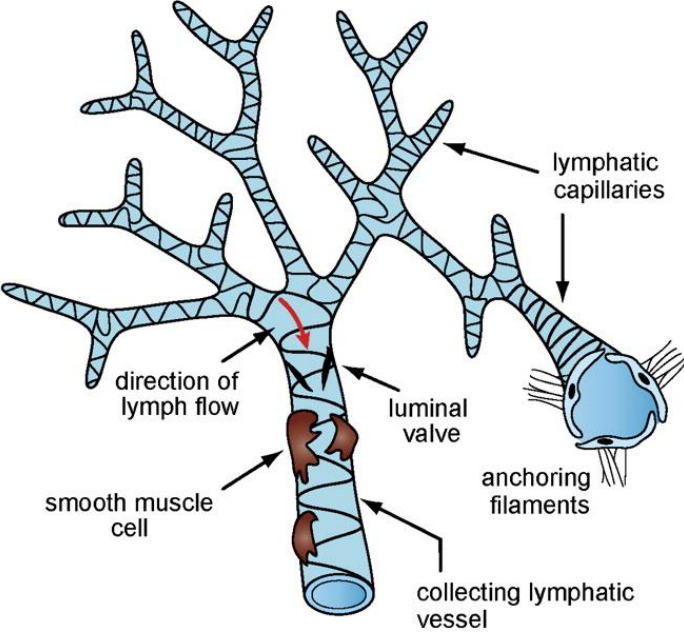
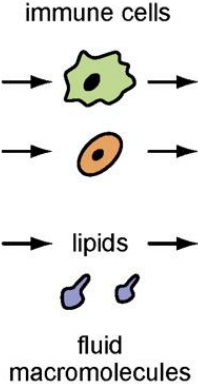
- Hippocrates 460–377 b.c.
lymphatic vessel: “white blood”
lymph (Latin): water
chylus (Greek): juice
- Gaspare Aselli 1627
re-identification lipid-filled
“milky veins” in the
gut of a “well-fed” dog
- Jean Pecquet 1651
lymphatic system in animals
- Thomas Bartolin 1652
lymphatic system in man



lymphatic system



Lymphatic vasculature



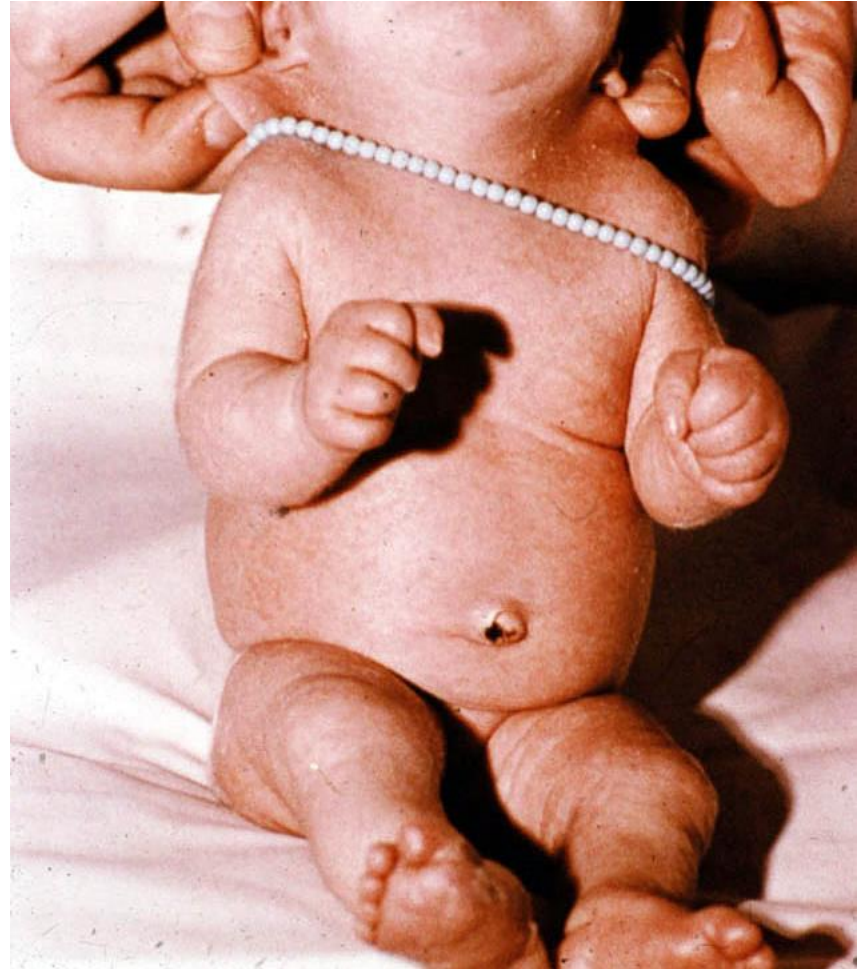


- circulation of fluid next to blood circulation
- functions
 - defence against infections
 - fluid household
 - fat absorption

lymphedema

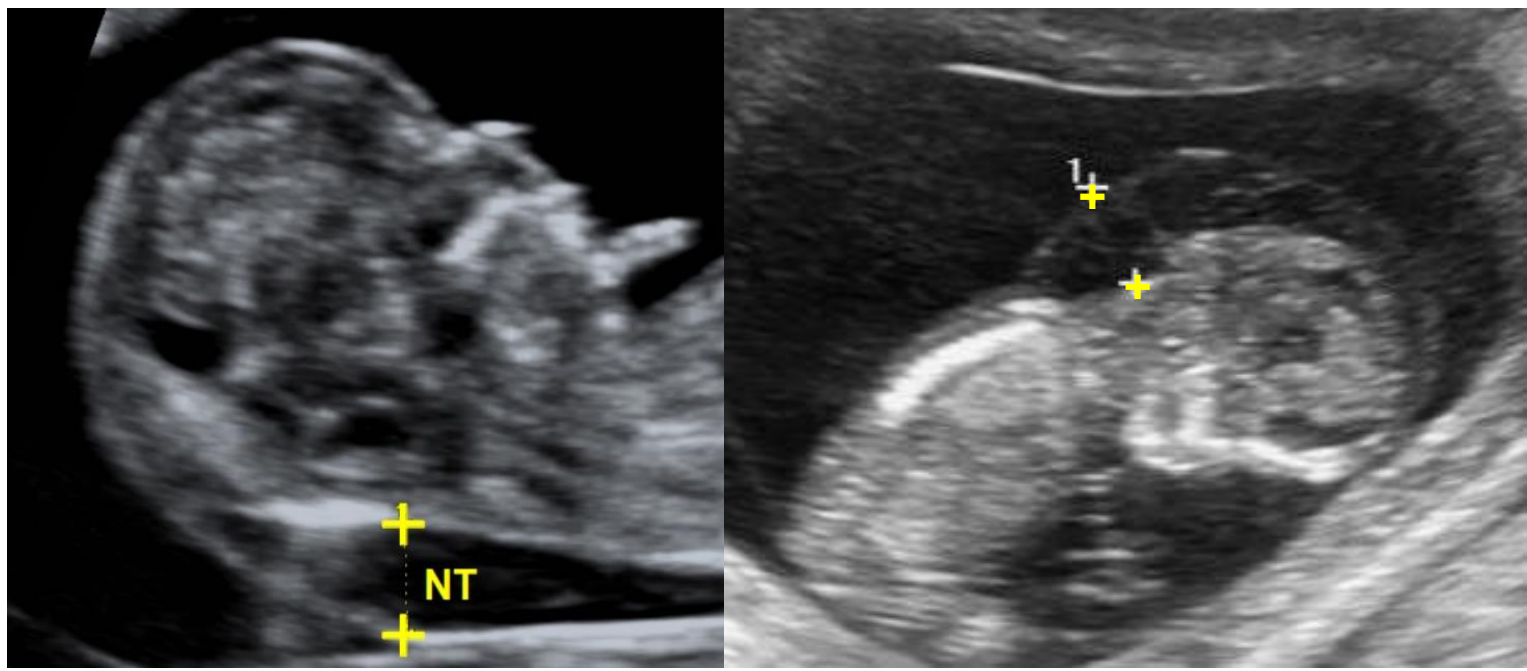


at birth





before birth



lymphedema



lymphedema



lymphedema



subtypes

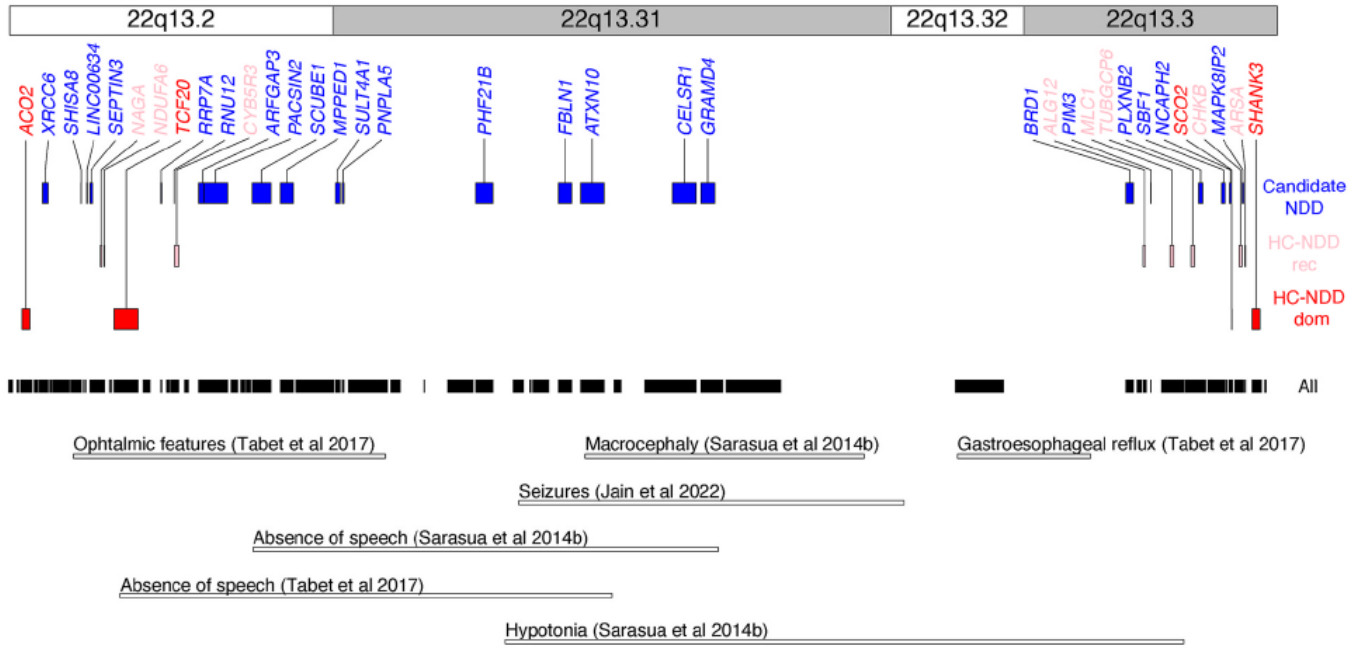


	<i>OMIM</i>	<i>Pubmed</i>
lymphedema	104	18747
primary lymphedema	43	866
lymphangiectasia	31	2142

search on October 2, 2023

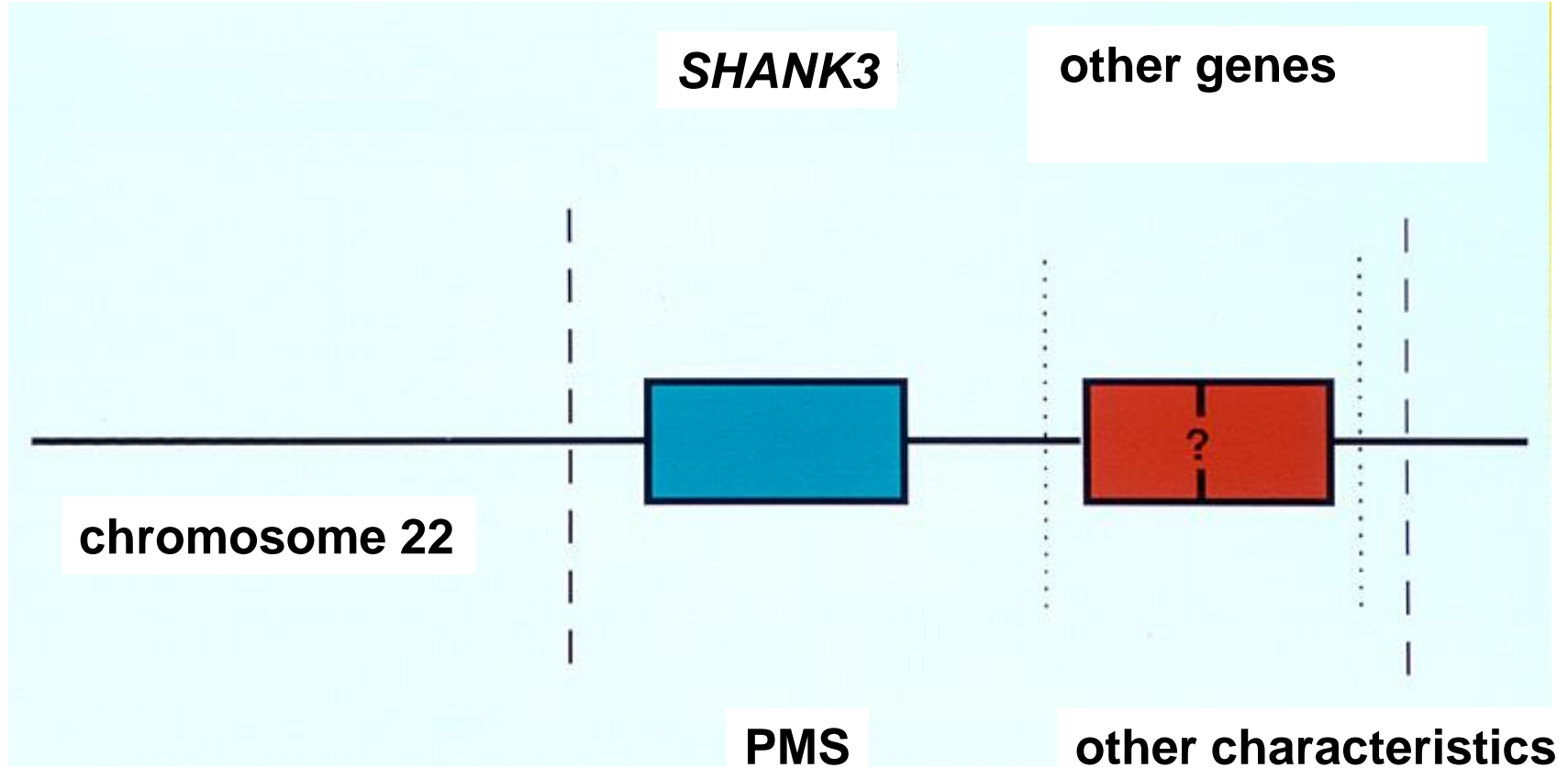


Phelan - McDermid syndrome





contiguous genes





lymphedema in PMS

- 11% in deletions
 - 0% in *SHANK3* variants
- birth or childhood
- progressive? (puberty)
- variable





lymphedema in PMS

- variable
- mainly feet
- sometimes hands
- rarely ascites / pleural effusion



management

- cure not possible
- physical activity
- avoid obesity
- compression
- skin care
- multidisciplinary / centres of expertise

ps https://vascern.eu/wp-content/uploads/2019/12/General-Patient-Pathway-for-PPL_08112019



Thank you!