lymphedema, hereditary I

Eponyms: Nonne-Milroy syndrome

Inheritance: autosomal dominant

Semeiological Synthesis: Cutaneous disorder, isolated defect. Lymphedema of the lower extremities, sometimes involving one foot or several toes, without pain, due to defect of the lymphatic drainage system. Occasionally development of lymphangiosarcoma in adulthood.

Group Signs:
- Super group: DERMATOLOGICAL DISORDERS
  - cutis, changes in appearance and/or features
    - isolated dermatological disorders
    - keratinisation defects
    - keratosis, hyperkeratosis, keratoderma, scaling cutis
    - subcutaneous changes
      - lymphedema, lymphoedema

- Super aggreg. GASTROINTESTINAL DISORDERS
  - liver dysfunctions, liver anomalies
    - ascites

- GENITAL DISORDERS
  - male genitilia, modifications not including ambiguity
    - hydrocele

- LABORATORY DATA
  - chromosomal assignment
    - chromosome 5q localization
  - gene, structural-functional anomalies
    - FLT4 (VEGFR3) PCL fms-related tyrosine kinase 4, gene chr.5q35.3
  - gene analysis-DNA analysis

- NEOPLASTIC DISEASES
  - cancer, genetic features
    - tumour susceptibility

- OTHERS
  - inheritance
    - inheritance, autosomal dominant

Super group: NEOPLASTIC DISORDERS
Super aggreg: tumour, susceptibility
Aggregations: isolated dermatological disorders

Differential diagnosis:
- 15300 congenital, generalized lymphedema
- 7740 distichiasis-lymphedema syndrome
- 15303 lymphedema-hypoparathyroidism syndrome
- 15310 lymphedema-microcephaly syndrome
- 15330 lymphedema-ptosis syndrome

Bibliography
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