Key Points for Clinical Practice

Dermatological Pathologies

Common Dermatology and Trisomy 21 : Part 2

Individuals with T21 are at higher risk of superficial skin infections compared to the general population.
The most common dermatophytoses in individuals with T21 are onychomycosis and athlete's foot:

- **Trisomy 21 is recognized as a predisposing factor for onychomycosis:**
  - The etiology is typically due to a dermatophyte infection, notably Trichophyton rubrum.
  - The prevalence increases with age as in the general population.
  - Childhood cases are not rare and can represent half of the cases in some series, with an overrepresentation of Trisomy 21 in rare very early forms (< 2 years).
  - Toenail involvement is highly predominant, and it is nearly always associated with athlete's foot.
  - Preventive hygiene measures are essential. The treatment must be tailored to the cognitive capacities and sensory particularities of individuals with T21. Oral terbinafine has been found to be effective in these patients.

- **Athlete's foot (Tinea Pedis) is a frequent skin condition in Trisomy 21:**
  - Frequently associated with onychomycosis, athlete's foot presents similarly as in the general population, with Trichophyton interdigitale and Trichophyton rubrum being the most common pathogens responsible for the infection.
  - Preventive hygiene measures are essential. Once the diagnosis has been made and the etiology has been determined by culture, treatment must be tailored to the cognitive abilities and sensory particularities of these patients.
  - We have no data on the efficacy of the different treatments in this population.

Livedo is commonly observed in individuals with T21:

- **It is typically:**
  - Livedo reticularis (small network) and physiological livedo (Cutis Marmorata);
  - Often present at birth;
  - Triggered by exposure to cold and likely related to autonomic dysfunction, which is common in Trisomy 21;
  - It disappears entirely with warming, and the only recommended treatment is cold avoidance.
- **It rarely persists with warming. It can be:**
  - Primary livedo reticularis: Arteriolar vasoconstriction mainly explains this type of livedo, and elevating the affected limbs often reduces it;
  - Idiopathic livedo reticularis: This form is permanent and does not improve with warming. It requires follow-up, as it may be an early manifestation of a secondary form.

- Livedo racemosa (large network): further evaluation is required to assess for an underlying pathology (e.g., Hashimoto's thyroiditis, COVID-19 infection).

Three perioral lesions are commonly encountered in individuals with T21: lip fissures (chapping), unspecified cheilitis, and angular cheilitis or Perleche (AC):

- These lesions have a higher prevalence than in the general population.
- Many factors can explain this high prevalence: Hypoplasia of the median facial mass, muscle hypotonia (orbicularis oris), mouth breathing and labial incontinence, deep wrinkles at the oral commissures, vitamin B (B12) and mineral (Iron and Zinc) deficiencies; reduced salivary secretion and higher frequency of superficial fungal infections. Also, the prevalence of diabetes, psychiatric disorders, and "bad habits" (frequent licking, biting, and sucking of the lips).
- Treatment of the lesions follows the same principles as for neurotypical patients. However, in cases of angular cheilitis, due to the increased prevalence of Candida albicans—which is twice as common as in the general population—first line topical antifungal therapy should be initiated promptly after obtaining a culture sample, without awaiting culture results.

Acanthosis nigricans is a common skin disorder in individuals with T21, particularly adolescents with obesity or diabetes. Acral forms are frequent.

- The treatment involves addressing the cause (obesity or diabetes), with regression of lesions in some cases.

La Mélanocytose dermique congénitale, que l'on ne doit plus appeler « tache mongoloïde » ne semble pas plus prévalente chez les enfants PT21 que dans la population générale.

- Sa persistance ou son extension au-delà des premières années de vie, les lésions >10 cm, multiples, ou siégeant sur le visage ou l'épaule devront être référées à un dermatologue pour rechercher une erreur innée du métabolisme.

Congenital dermal melanocytosis, which should no longer be called "mongoloid spot", does not appear to be more prevalent in children with T21 than in the general population.

- Persistence or extension beyond the first few years of life, lesions >10 cm, multiple lesions, or lesions on the face or shoulders should be referred to a dermatologist to evaluate for inborn errors of metabolism.

Café-au-lait macules (CALMs) do not appear to be more prevalent in children with T21 than in the general population.

- The vast majority of CALMs described in Trisomy 21 are isolated, and reported cases of CALMs with a genetic syndrome, notably Neurofibromatosis type 1 (NF1), are rare and probably anecdotal.
- However, due to reported cases of the double diagnosis of Trisomy 21 + NF1, vigilance is required, and the patient should be referred to a dermatologist in cases of large and multiple CALMs or atypical localization, notably axillary or inguinal freckling.