NOTES
PIGMENTATION DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Skin loses pigment, becomes lighter/darker
- Pigment-producing cells (melanocytes)
  - Expected number, overproduce pigment (melanin)
  - Higher in number, produce expected melanin
  - Destroyed, no melanin produced

CAUSES

- Autoimmune disorders → melanocyte destruction
- Long periods of sun exposure
- Genetic mutations

SIGNS & SYMPTOMS

- Changes in skin pigmentation only symptom
- Some disorders (esp. produced by sun overexposure) heighten risk of skin cancer

DIAGNOSIS

LAB RESULTS

- Skin biopsy
- Genetic testing

OTHER DIAGNOSTICS

- Dermatological physical examination (e.g. dermoscopy)

TREATMENT

- Usually no cure

MEDICATIONS

- Topical medication
  - Most respond to steroids
  - Depigmentation agents effective for hyperpigmentation disorders

SURGERY

- For some lesions

OTHER INTERVENTIONS

- Sun avoidance, if caused by ultraviolet (UV) overexposure

ALBINISM

osms.it/albinism

PATHOLOGY & CAUSES

- Congenital condition; skin, eyes, hair partially (hypomelanistic albinism)/ completely (amelanistic albinism) devoid of pigment
- Lack of pigment → dermatological problems
  - Increased risk of sunburn, skin cancer

TYPES

Oculocutaneous albinism (OCA)

- Eyes, skin; possibly hair
- Autosomal recessive transmission
- Seven different subtypes; OCA1, OCA2 most common
  - OCA1: defect in gene for enzyme tyrosinase (TYR)
  - OCA2: defect in P gene (membrane transporter, moves amino acid tyrosine)
- Rufous oculocutaneous albinism
  - Specific subtype of OCA
  - Common in people of sub-Saharan African descent

Ocular albinism (OA)

- Only eyes
- OA1 most common subtype (AKA Nettleship–Falls syndrome)
  - X-linked recessive inheritance
  - Lack of pigment in retinal epithelium

CAUSES

- Hereditary/genetic
  - Autosomal/X-linked
  - Recessive inheritance pattern → defect leading to lack/absence of enzyme in melanin synthesis pathway → hypopigmentation/depigmentation
- Chediak–Higashi syndrome
  - Rare; malfunctions in lysosomal trafficking regulator gene (*CHS1/LYST*)

SIGNS & SYMPTOMS

- Complete/partial absence of skin pigmentation
- Light yellow/white hair
- Light eye colour
  - Light blue (partial pigment production)
  - Pink (complete absence of pigment production)
- Visual problems
  - Visual development in fetus highly dependent on melanin production; abnormal arrangements in optic nerves fibres (e.g. abnormal optic chiasm)
  - Severe sensitivity to light
  - Poor visual acuity due to foveal hypoplasia
  - *Amblyopia/nystagmus:* poor coordination between eye, brain

DIAGNOSIS

LAB RESULTS

- Genetic testing
  - Identify defective gene, allotype

OTHER DIAGNOSTICS

- Physical examination
- Family history

TREATMENT

- No cure

SURGERY

- Manage strabismus, nystagmus

OTHER INTERVENTIONS

- Lifestyle management
  - Avoid sunburn
  - Regular dermatological, ophthalmological check-ups
  - Visual rehabilitation
  - Glasses/contact lenses

Figure 7.1 A baby with albinism.

PITYRIASIS ALBA

osms.it/pityriasis-alba

PATHOLOGY & CAUSES

- Common, irregularly hypopigmented skin condition: slightly scaly patches, macules
  - *Lesion diameter:* 0.5–5cm/0.2–2in
  - Irregular borders
  - Most common on face, neck, shoulders, upper arms
  - May resolve spontaneously after puberty

CAUSES

- *Unknown etiology:* eczema-related post-inflammatory hypopigmentation (possible relation)

RISK FACTORS

- Atopy
- Sun exposure
- Frequent bathing
- Biologically male
- Children/adolescents > adults

COMPLICATIONS

- Benign condition

SIGNS & SYMPTOMS

- Often asymptomatic, may itch/burn

DIAGNOSIS

OTHER DIAGNOSTICS

- *Wood's lamp examination:* hypomelanosis patches
- *Microscopy:* ↓ melanocyte number, size

TREATMENT

MEDICATIONS

- Hydrocortisone (topical), calcineurin inhibitors, emollients

VITILIGO

osms.it/vitiligo

PATHOLOGY & CAUSES

- Pigmentation disorder; parts of skin, hair lose pigment
- Melanocytes destroyed → white patches of depigmented skin
  - Sharp margins on depigmented patches
- May be autoimmune condition

TYPES

Segmental

- Areas innervated by dorsal roots of spinal cord
- Unilateral; stable over time

Non-segmental

- Symmetrical; appearance of new patches
- Multiple subtypes
  - *Vitiligo universalis:* most severe, almost no pigmented skin remains
  - *Generalized:* most common
  - *Focal:* smaller, localised patches
  - *Acrofacial:* hands, face
  - *Mucosal:* only mucous membranes

CAUSES

- Autoimmune disorder → melanocyte destruction

RISK FACTORS

- Medical/family history of autoimmune conditions
  - Hashimoto's thyroiditis
  - Type I diabetes mellitus
  - Systemic lupus erythematosus
  - Celiac disease
  - Addison's disease

SIGNS & SYMPTOMS

- Depigmented skin patches only symptom, usually on extremities
- Patches grow over time (non-segmental subtype)
- Can be associated with alopecia

DIAGNOSIS

- Rule out autoimmune/inflammatory disorders

OTHER DIAGNOSTICS

- *Ultraviolet light (Wood's lamp):* lesions; vitiligo turns blue

TREATMENT

- No cure

MEDICATIONS

- Topical immune system suppressing medication
- Glucocorticoids
- Calcineurin inhibitors

OTHER INTERVENTIONS

- Ultraviolet light therapy (phototherapy)
- Skin camouflage (e.g. makeup)

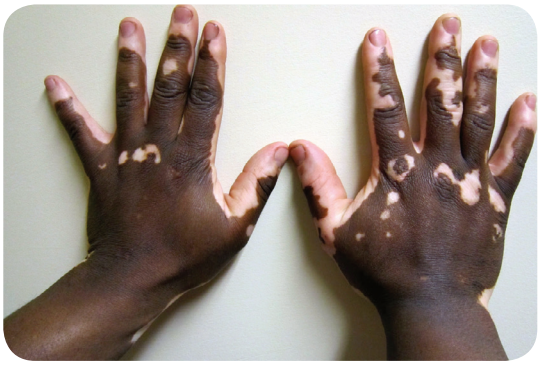

Figure 7.2 The clinical appearance of vitiligo affecting the hands.