Mastocytoses Systémiques: Diagnostic et Prise en charge

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19 mars 2016

Hôpital Erasme

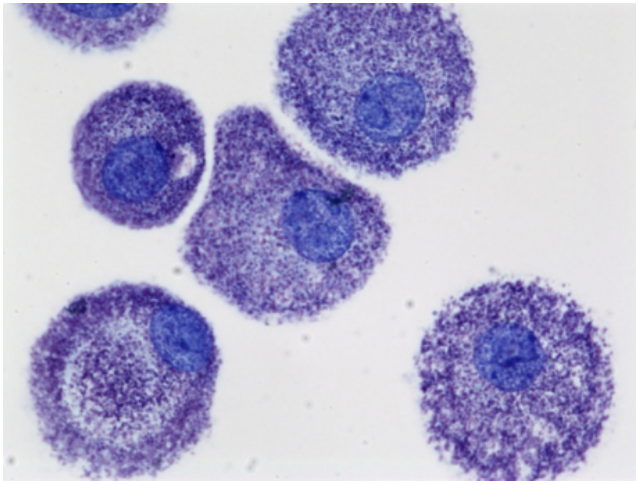
ULB

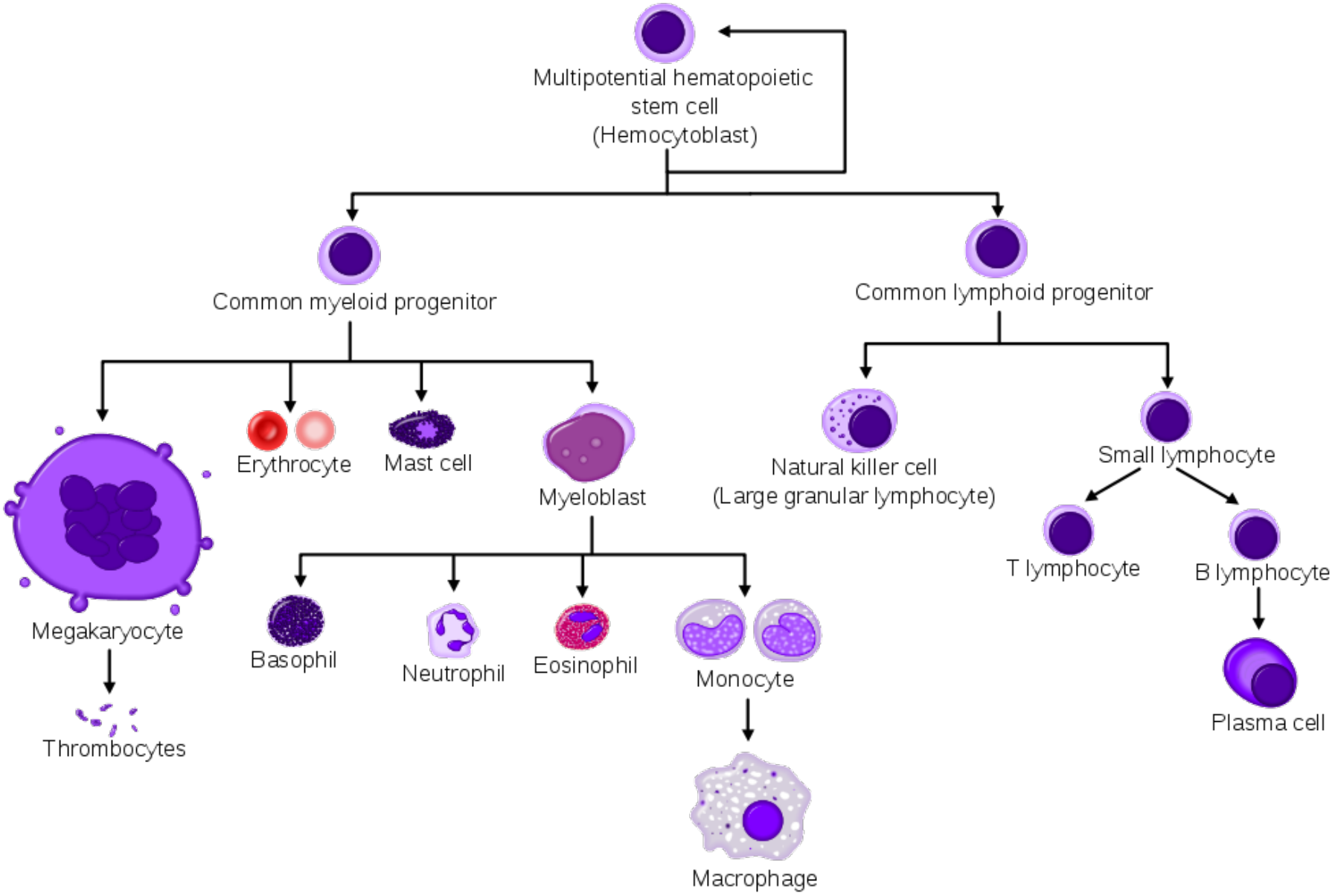
CHU tivoli

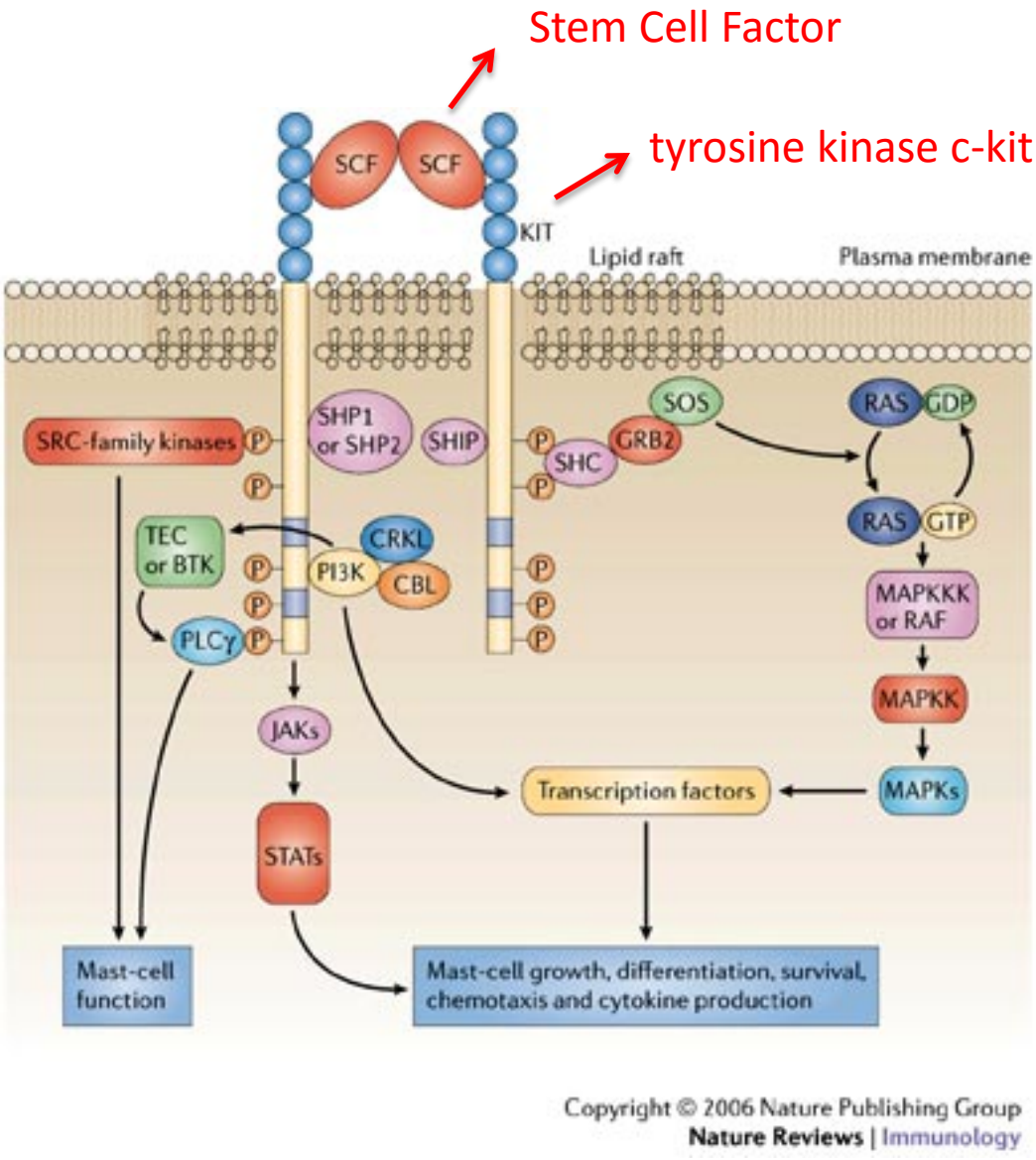
Les Mastocytoses

- Groupe hétérogène de pathologies acquises, caractérisées par une **prolifération** et **accumulation anormales de mastocytes** dans différents tissus.

- Peau → Mastocytose Cutanée
- Organes extra-cutanés → Mastocytose Systémique

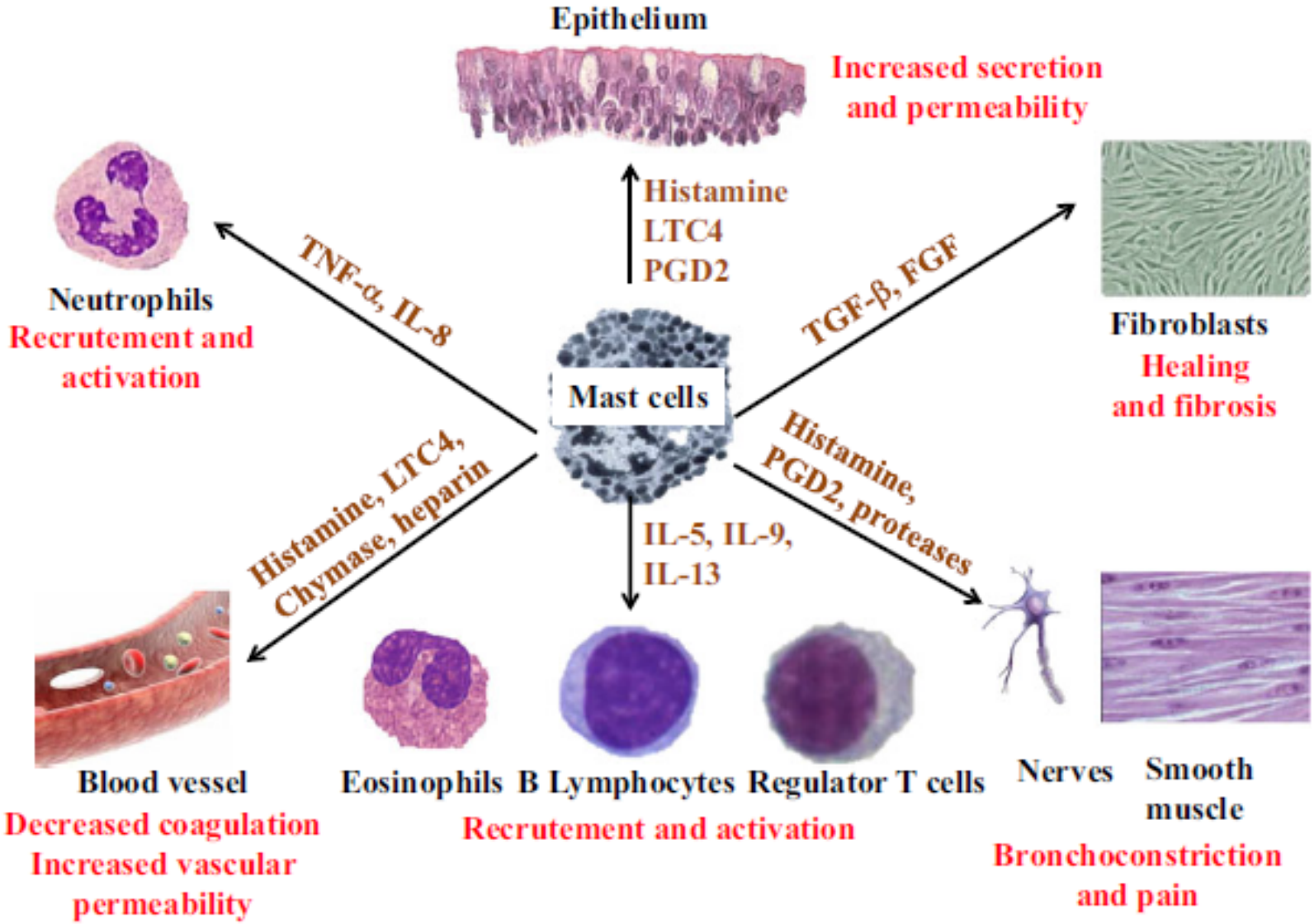



Multipotential hematopoietic stem cell (Hemocytoblast)
Common myeloid progenitor
Common lymphoid progenitor
Megakaryocyte
Thrombocytes
Erythrocyte
Mast cell
Myeloblast
Basophil
Neutrophil
Eosinophil
Monocyte
Macrophage
Natural killer cell (Large granular lymphocyte)
Small lymphocyte
T lymphocyte
B lymphocyte
Plasma cell



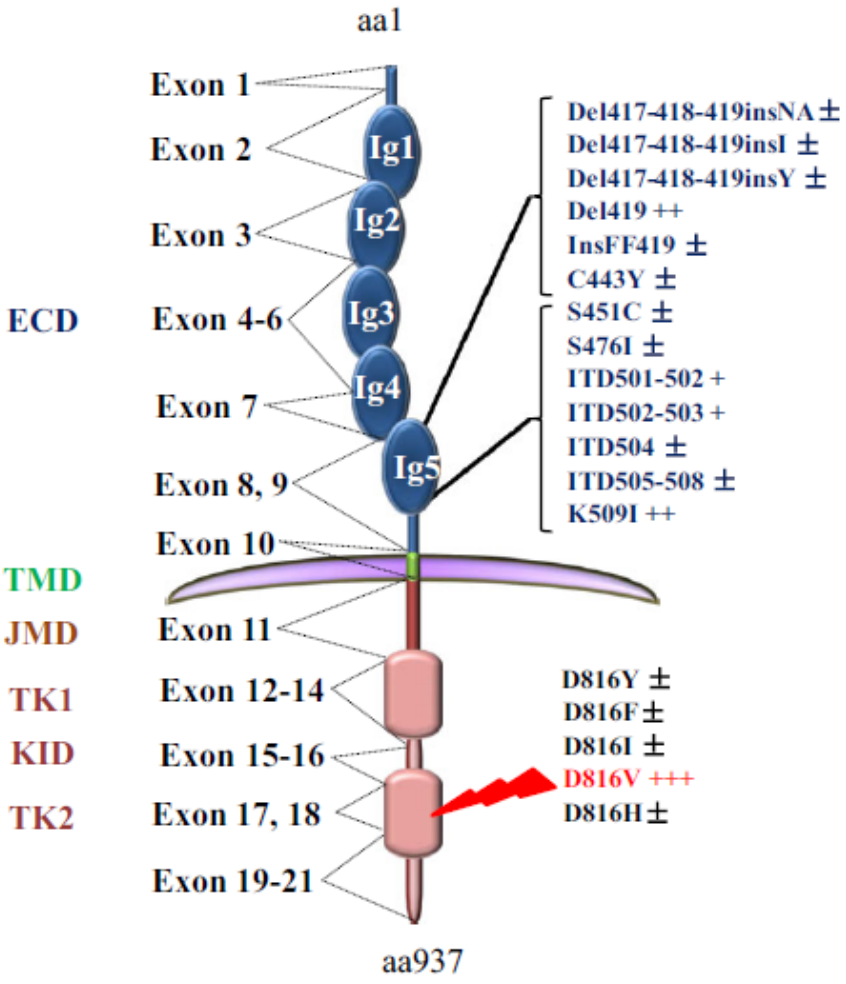
- Peau
- Muqueuses respiratoires, digestives, urogénitales
- Paroi des vaisseaux

Peu dans les reins,
Peu dans la moelle osseuse



Arock et al, Eur J Hematol 2015

Mastocytoses: Mutation gène KIT



Mutation du gène codant pour le récepteur KIT (D816V +++) (gain de fonction)

↓

Expansion clonale et défaut d'apoptose

↓

Accumulation pathologique de mastocytes au niveau des tissus

Arock et al, Eur J Hematol 2015

Epidémiologie

- Maladie rare, incidence exacte inconnue
- En Europe, la prévalence est estimée à 1 – 1.3 cas pour 10.000 habitants
- Homme = Femme
- Chez l'enfant, 80% avant 1 an, surtout formes cutanées
  En général régression spontanée avant adolescence
- Chez l'adulte, le plus souvent formes systémiques (95%)

Bonadonna et al, J Investig Allergol Clin Immunol 2014

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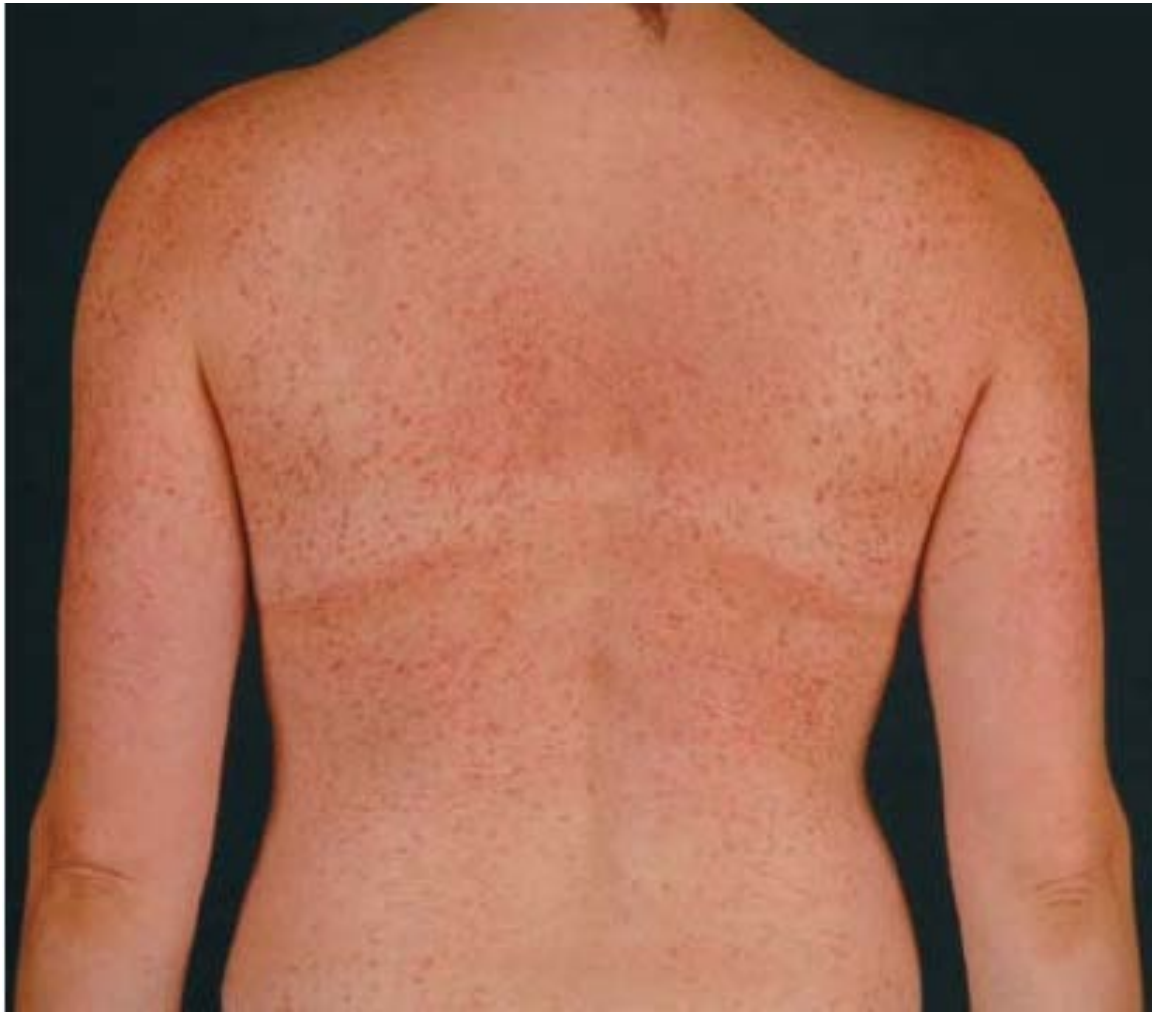
Urticaire pigmentaire



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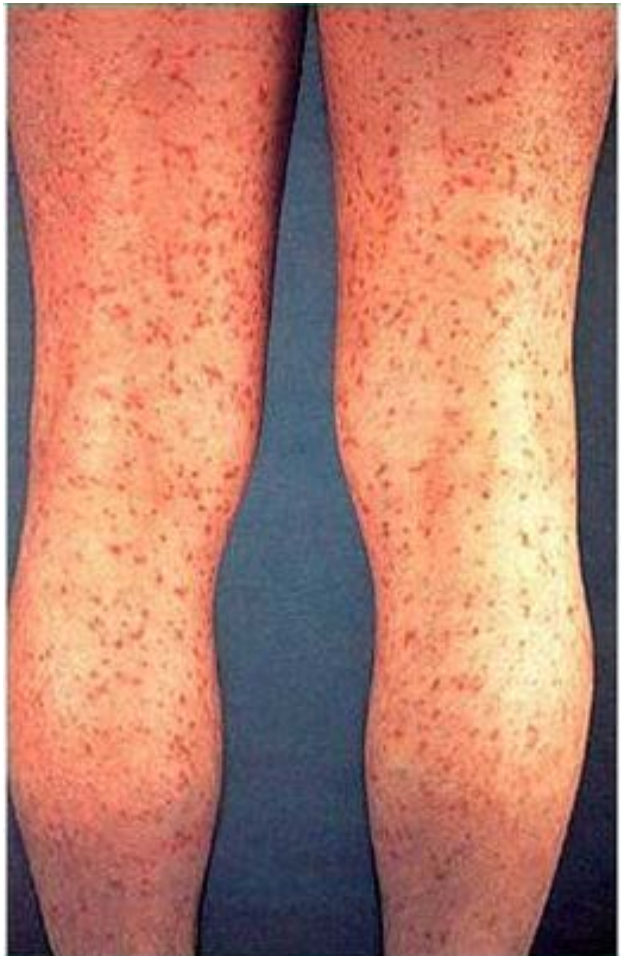
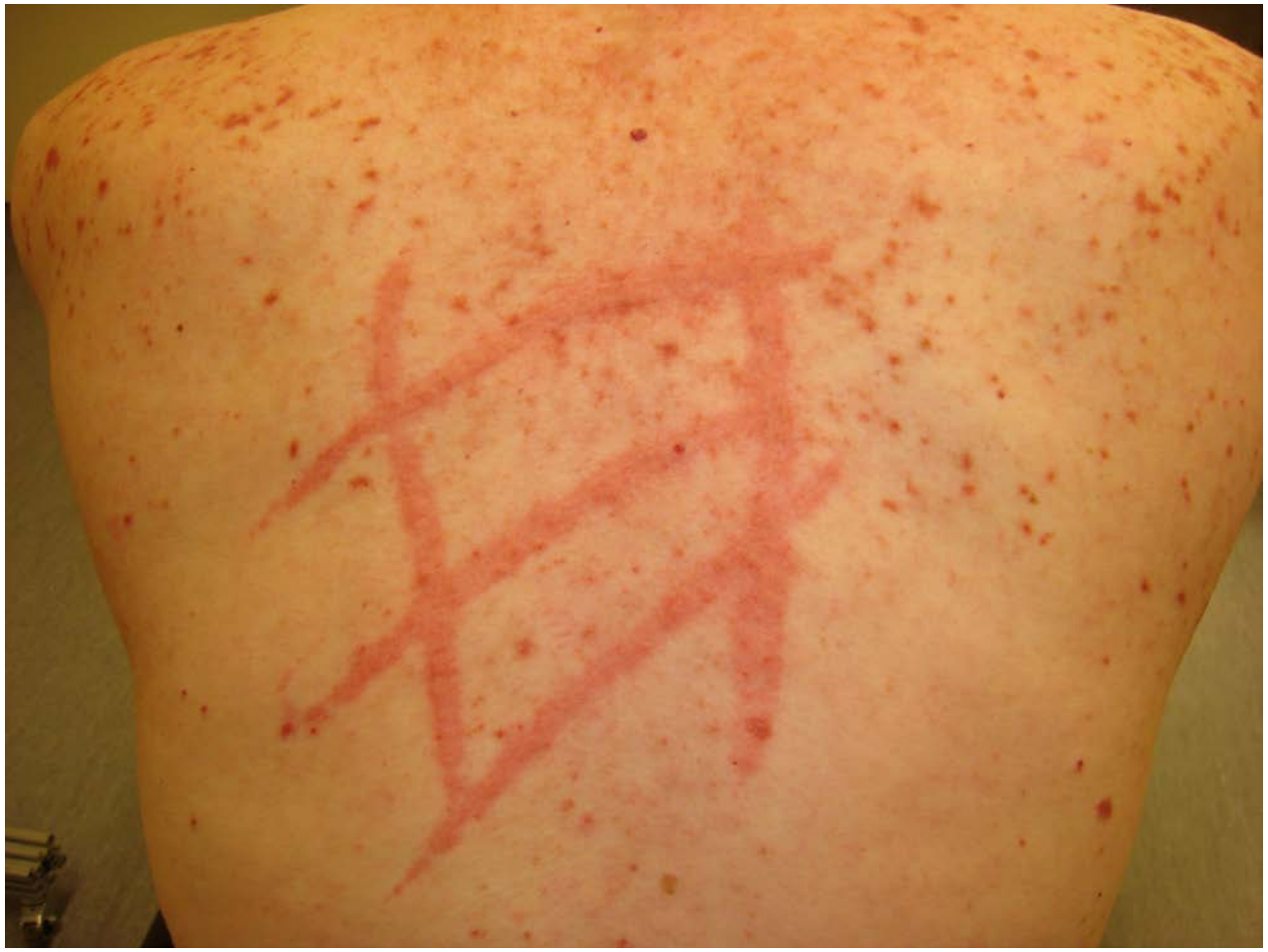


Signe de Darier



Prurit et UP exacerbé par certains facteurs

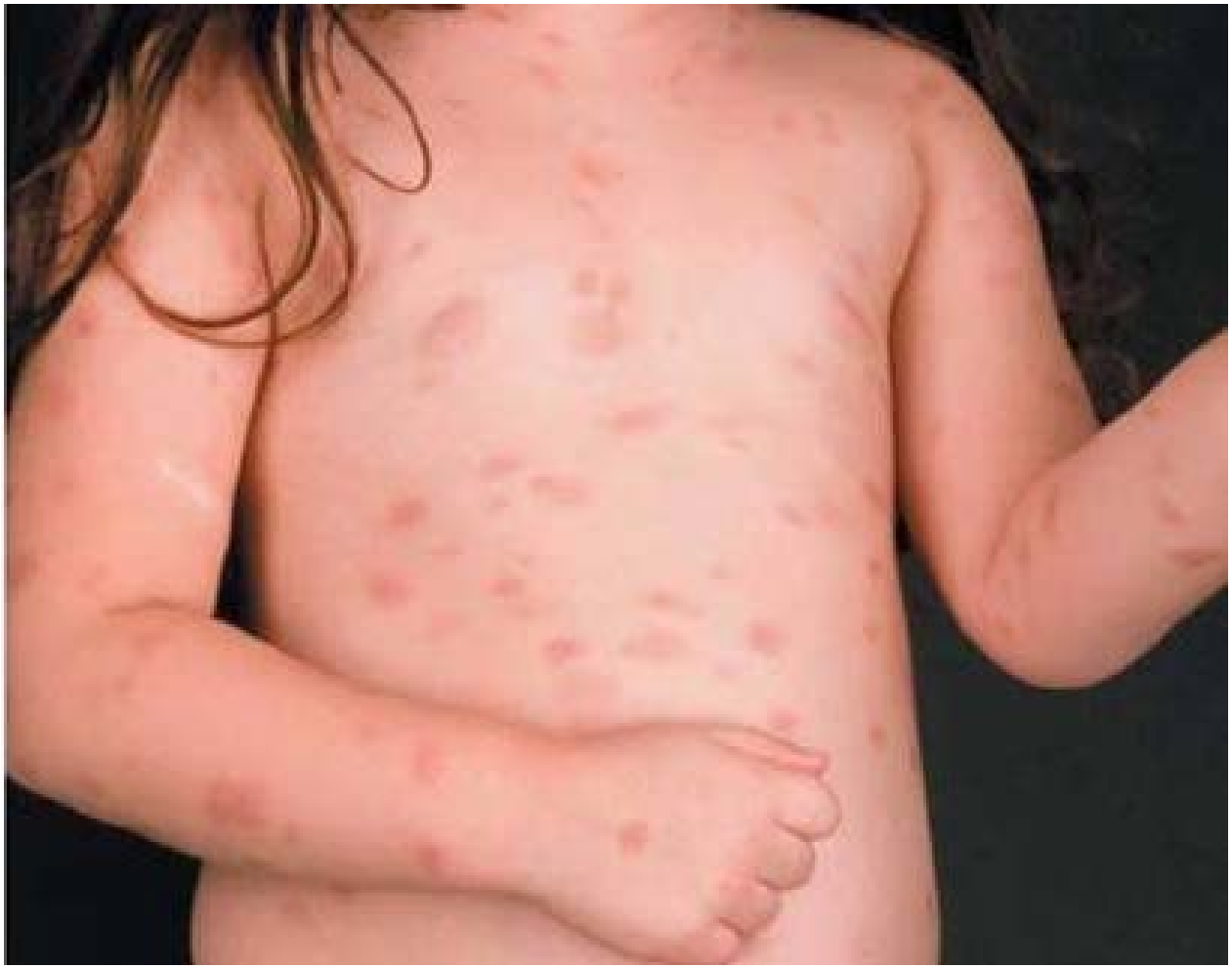
Facteurs déclenchants

Encadré 1. Facteurs pouvant favoriser la dégranulation mastocytaire.

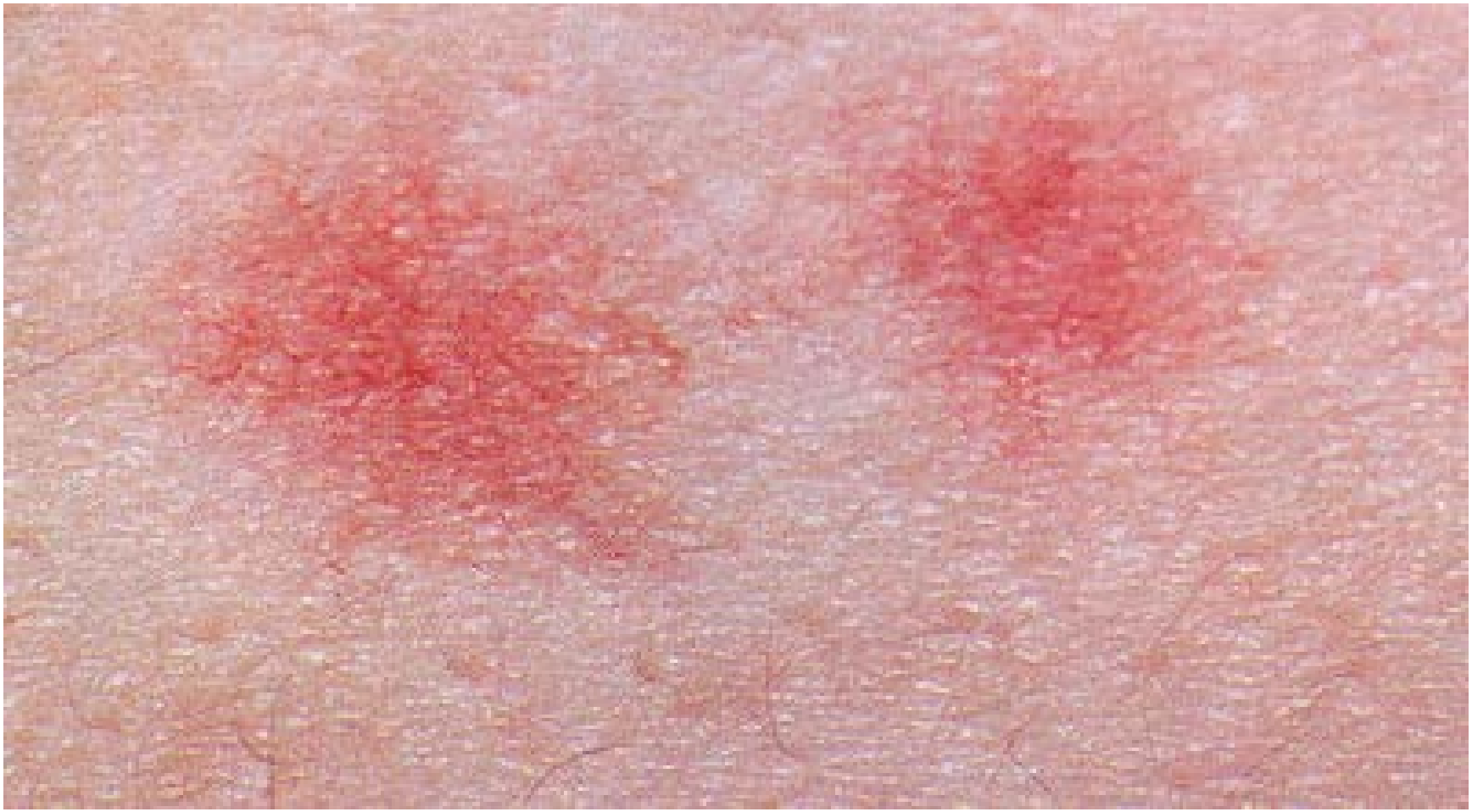
Variations thermiques marquées (bains chauds)
Exercice physique intense, traumatismes
Émotions et stress
Venins d'hyménoptères (abeille, guêpe)
Aliments histamino-libérateurs : alcool, œufs, chocolat, fraises, ananas, fruits exotiques, crustacés, poissons, tomates...
Médicaments et apparentés : aspirine (indiquée cependant dans certains cas), anti-inflammatoires non stéroïdiens, anticholinergiques, myorelaxants, opiacés, codéine, codéthyline, procaïne, lidocaïne, polymyxine B, amphotéricine B, quinine, réserpine, hydralazine, pentazocine, thiamine, interféron alpha (indiqué cependant dans certains cas), dextran, mannitol, produits de contraste iodés

Barete S. Ann Derm Vener 2014

UP: forme nodulaire



UP: forme télangiectasique



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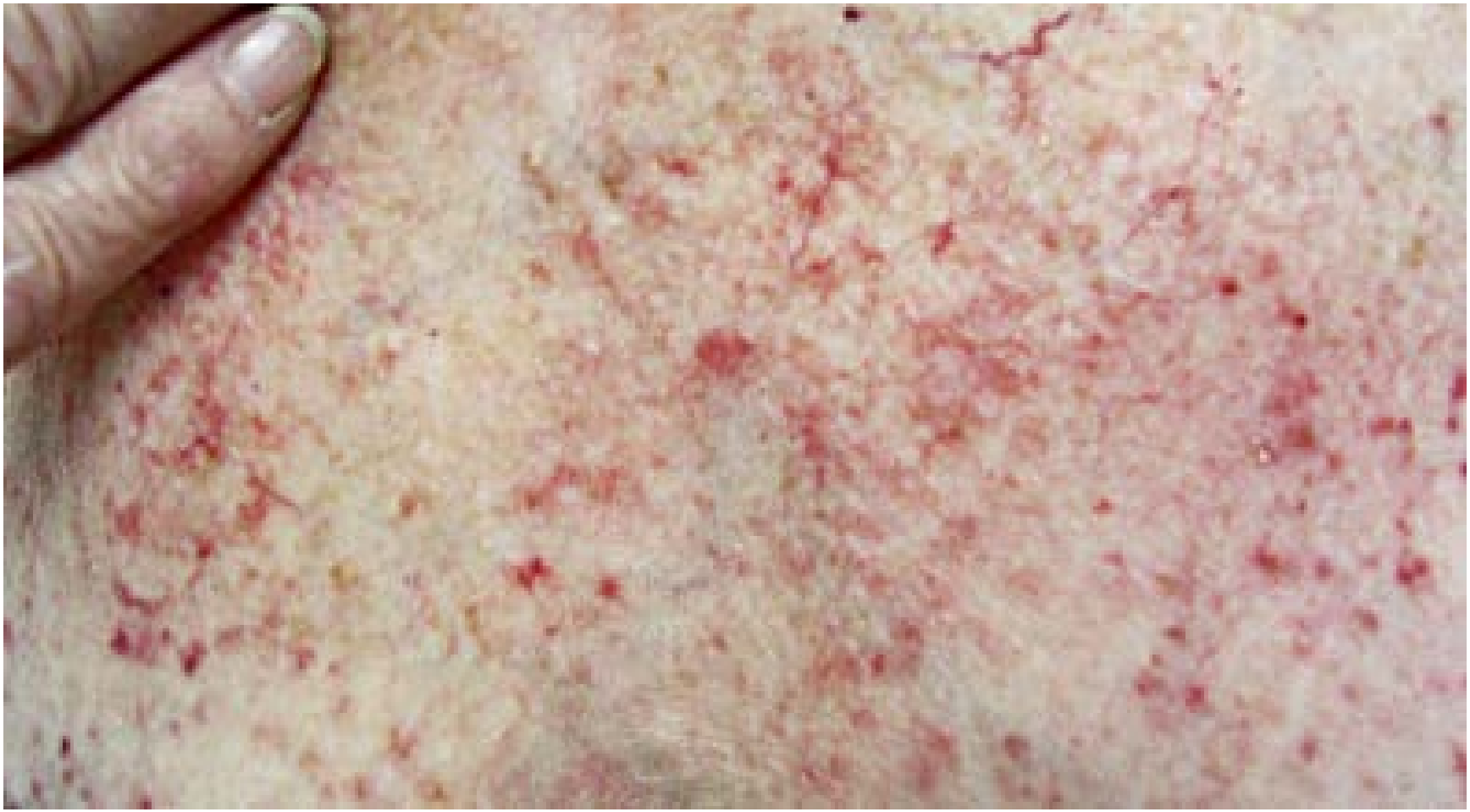

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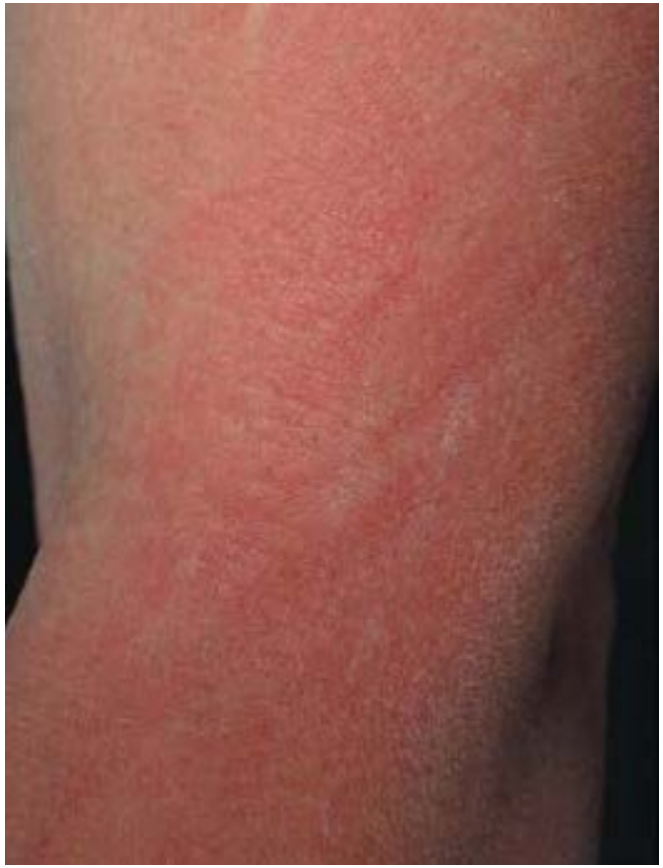
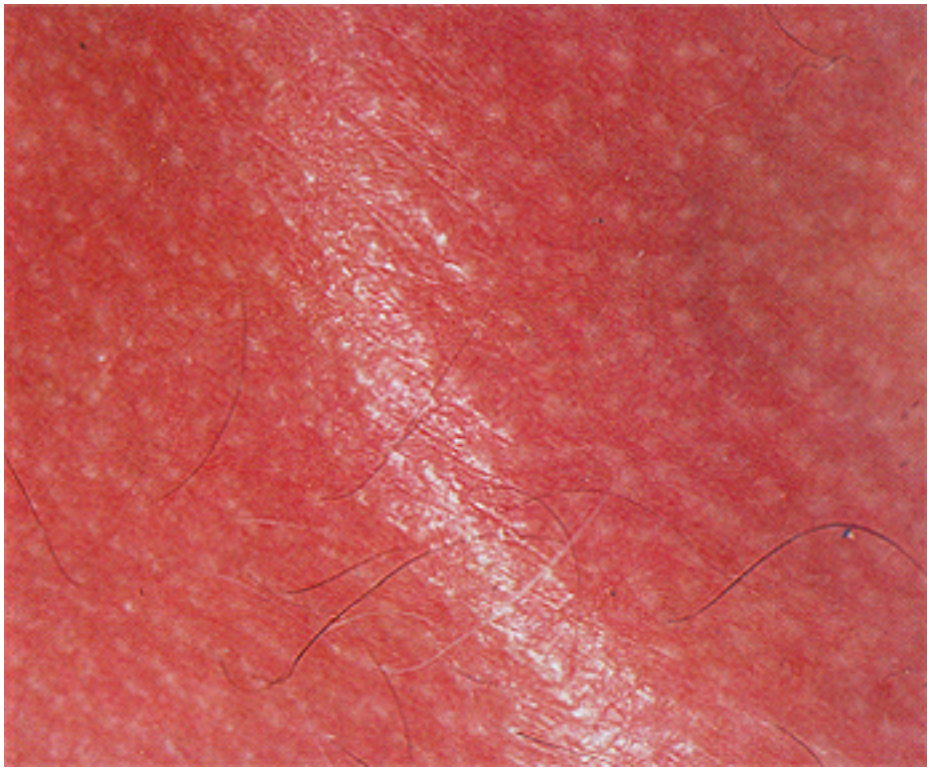
Mastocytose cutanée diffuse





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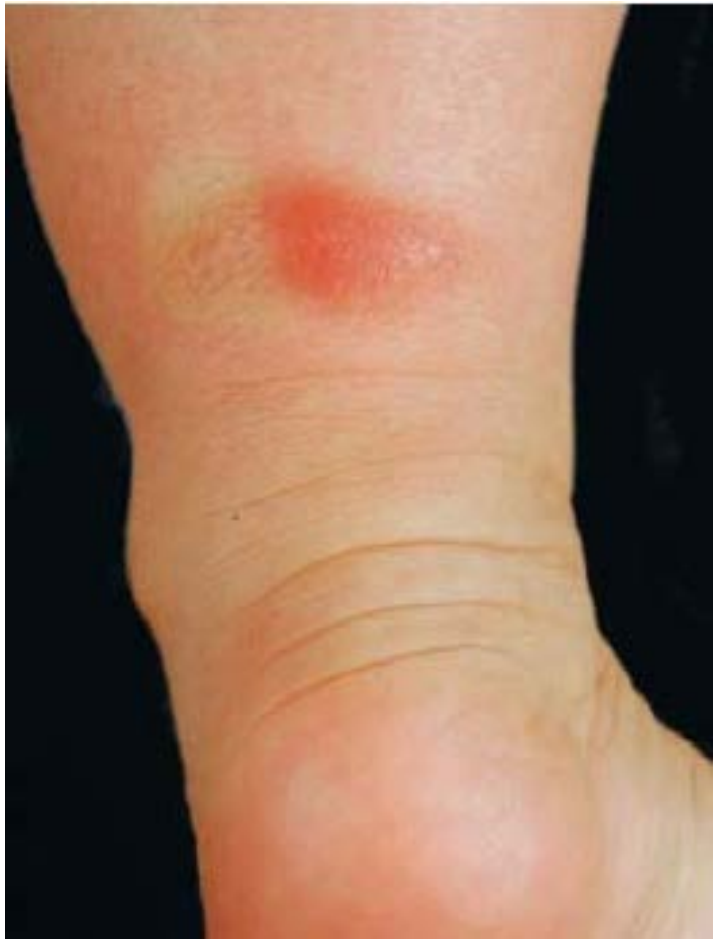
Mastocytomes de la peau



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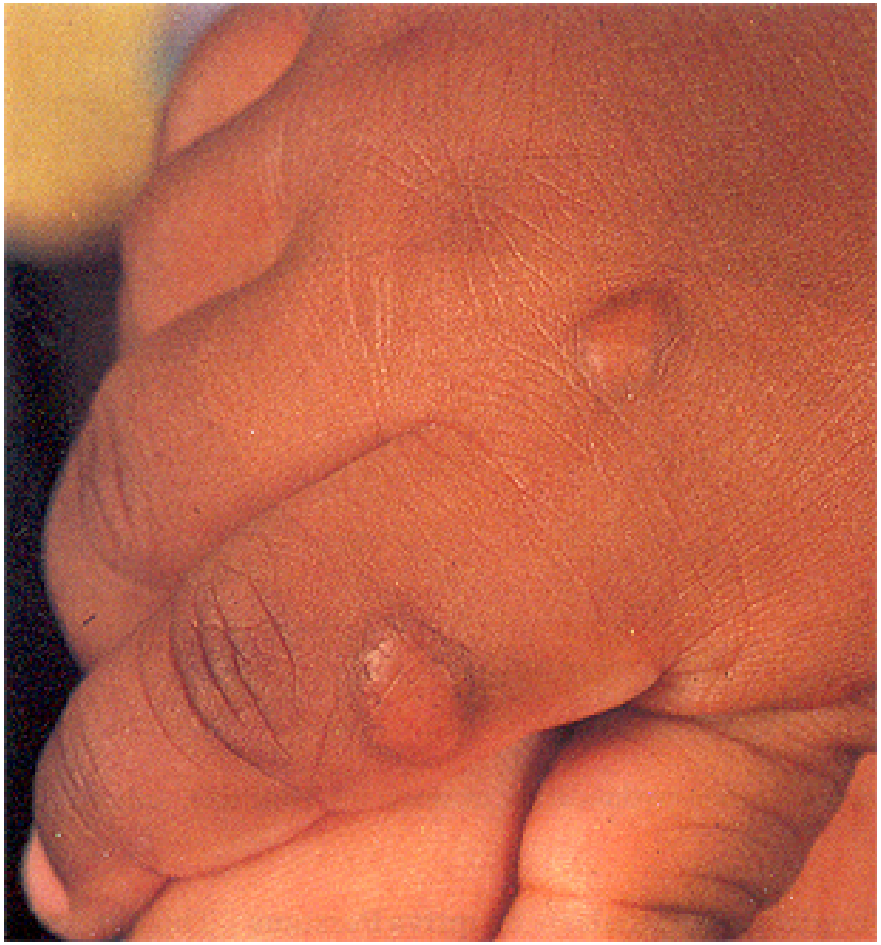


Manifestations Cliniques

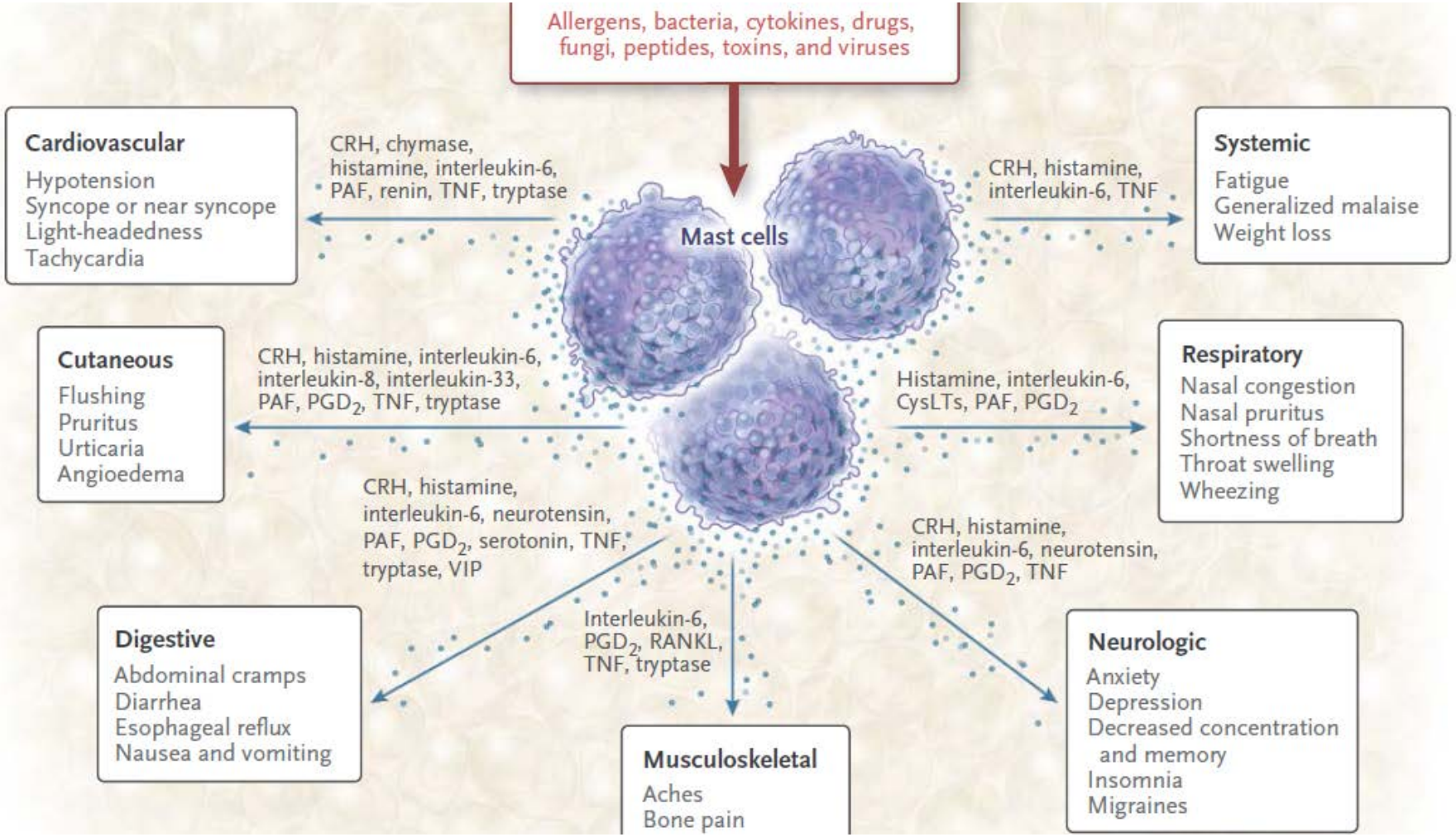
- Lésions cutanées:

  - Urticaire pigmentaire ++
  - Mastocytomes de la peau
  - Mastocytose cutanée diffuse

- Symptômes liés à la libération de médiateurs

- Symptômes liés à l'infiltration d'organes extra-cutanés (Mastocytoses Systémiques uniquement)

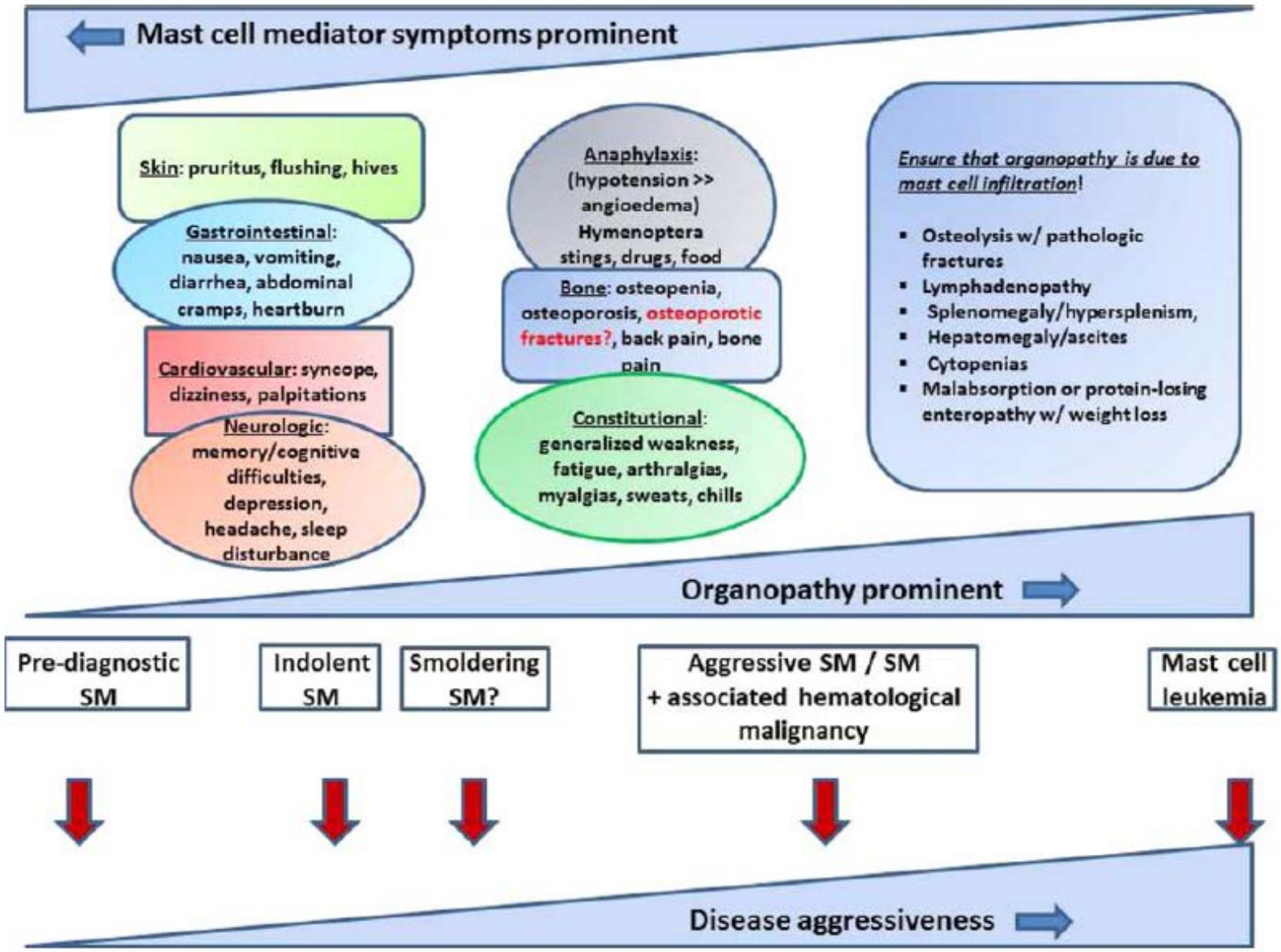
Symptômes liés à la libération de médiateurs



Theoharides et al. N Eng J Med 2015

Symptômes liés à l'infiltration d'organes

Moelle osseuse	anémie (50%), eosinophilie (25%), monocytose
Ganglions	adénopathies
Foie	HM,altération tests hépatiques, HTP, ascite
Rate	splénomégalie
Tube digestif	ulcère gastrique, malabsorption
Tractus génito-urinaire	pollakiurie
Squelette	lésions lytiques osseuses, fractures pathologiques (37% dans MSI)



Pardanani et al, Am J Hematol 2015

Diagnostic

- Sous-diagnostiqué

- Si lésions cutanées → biopsie cutanée

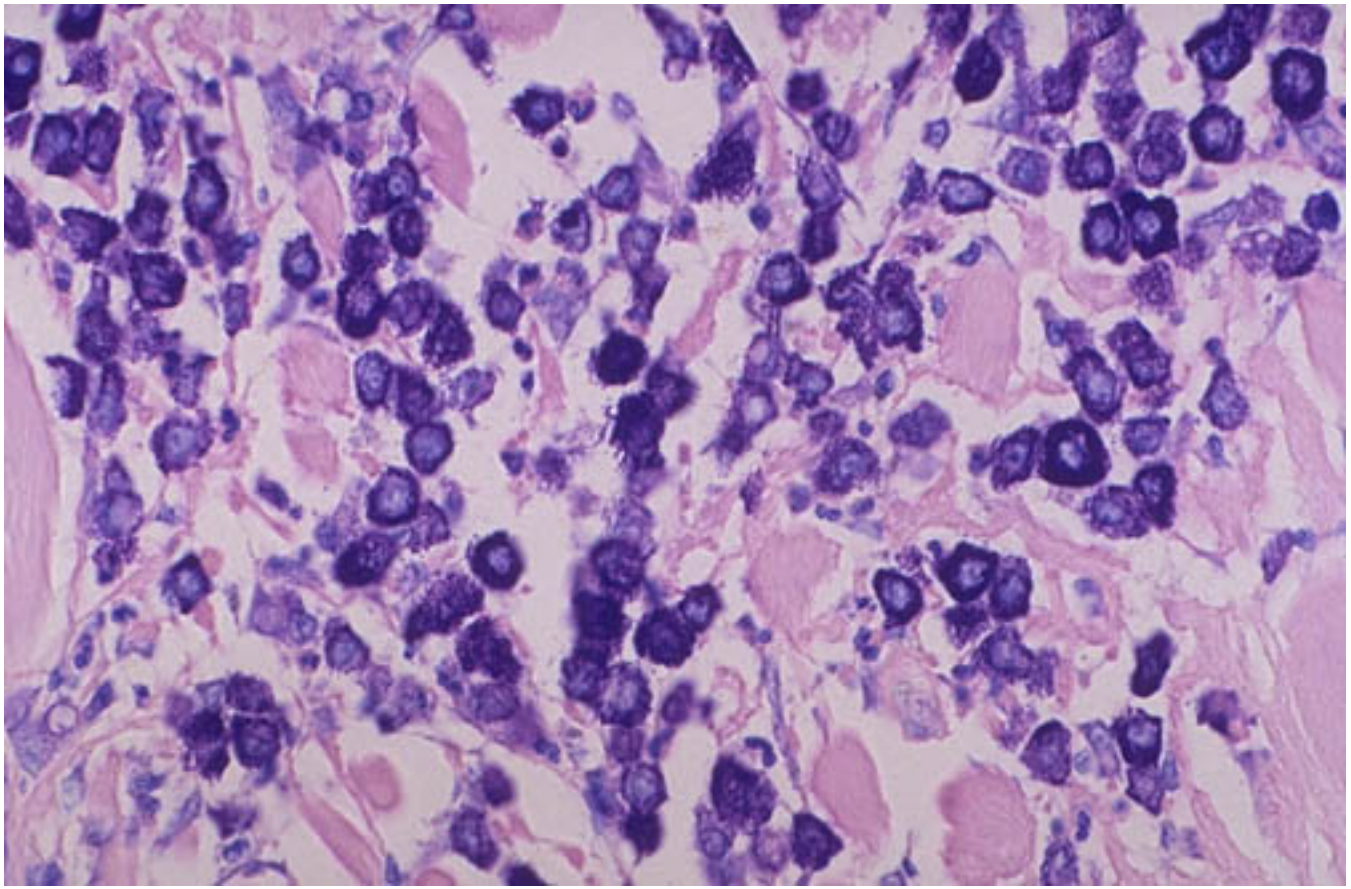
  Si + → Bilan de MS d'office chez l'adulte

- Si pas de lésions cutanées → y penser en cas de:

  - anaphylaxie inexpliquée ou récurrente
  - flush
  - ostéoporose
  - crampes abdominales récurrentes

  → bilan de MS

MC: Biopsie cutanée



- Coloration Giemsa
- Immunohistochimie: anticorps anti-tryptase
- Mastocytes parfois atypiques

Bilan de MS

- Prise de sang
- Ponction(-biopsie) médullaire
- Recherche mutation KIT

- Autres examens selon la clinique:
  - Rx de squelette des os longs,
  - CT thoraco-abdominal,
  - Gastro- ou colonoscopie,
  - Ostéodensitométrie osseuse
  - Echo abdominale

Biologie

- Biologie complète (hémogramme, enzymes hépatiques)

- **Tryptase sérique**

  LE marqueur des mastocytes

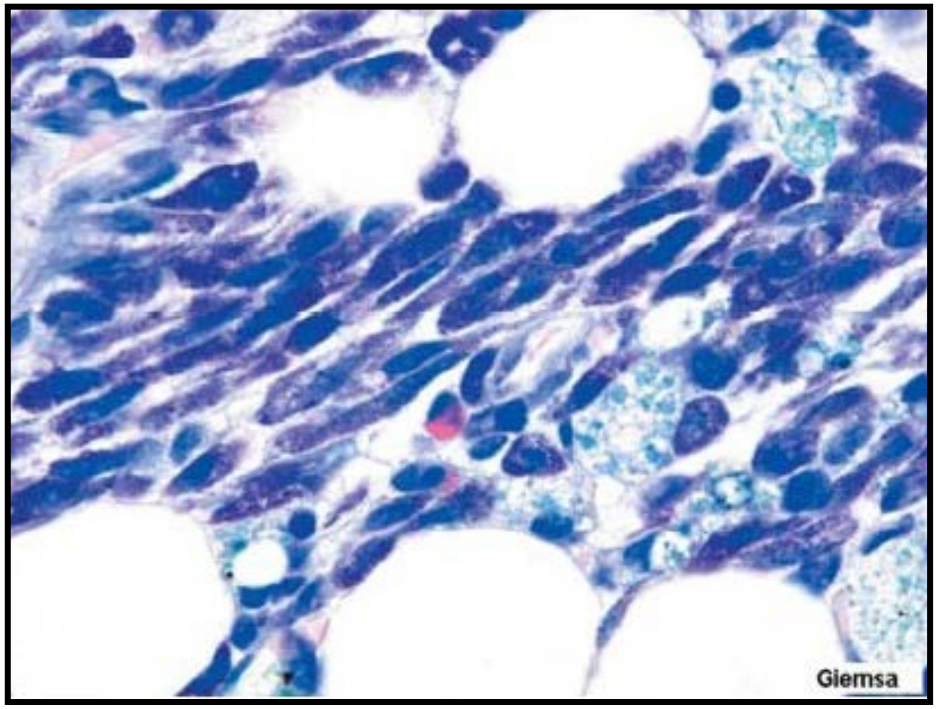
  N: 1 – 11.4 ng/ml

  Si > 20 ng/ml: suspicion +++ de MS

  ! SMD ou SMP, IRC ou IH, Urticaire chronique

Biopsie ostéo-médullaire

- Habituellement mastocytes rares (< 0.1% des cellules nucléées)
- MS: infiltrats ou agrégats multifocaux de > 15 mastocytes atypiques



Phénotype typique:
- CD117+ (récepteur c-kit)
- CD25+ et/ou CD2+
- tryptase
- chymase

Recherche de mutation KIT

Table 3 KIT mutants detectable in patients with mastocytosis

KIT mutant	Mastocytosis variant (estimated frequency)	Other non mast cell-lineage disorders
D816V	SM (70%–90%), CM (10%–30%)	GIST (< 1%), AML (< 5%), germ cell tumours (10%–20%)
D816Y	SM (< 5%), CM (< 5%)	
D816F*	SM (< 5%), CM (< 5%)	
D816H	SM (< 5%)	germ cell tumours (5%–10%)
R815K*	CM (< 1%)	
I817V	SM (< 1%)	
D820G	SM (< 1%)	
E839K*	CM (< 1%)	
V533D*	CM (< 1%)	
V560G	SM (< 1%)	GIST (< 5%)
V559A*	CM (< 1%)	
F522C†	SM (< 1%)	
del419†	SM-familial type (< 1%)	GIST-familial type
K509I†	CM/SM-familial type (< 1%)	
A533D*†	CM-familial type (< 1%)	

*So far only reported in children; †described as germ line mutations; GIST, gastrointestinal stromal cell tumour.

Sur la moelle
(éventuellement sur le sang ou la peau)

+ FIP1L1-PDGFRa si éosinophiles augmentés
+ JAK2 et bcr-abl si hyperleucocytose

Valent et al, Eur J Clin Invest 2007

MS: Critères diagnostiques de l'OMS

- Critère Majeur:

  présence d'agrégats de >15 mastocytes (dans moelle ou autre organe extra-cutané)

- Critères Mineurs:

1. morphologie anormale de > 25% des mastocytes
2. mutation KIT D816V
3. mastocytes CD2+ et/ou CD25+
4. tryptase sérique > 20 ng/ml

Majeur + 1 mineur OU 3 mineurs

Valent et al, Leuk Res 2001

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2 variantes:
- « smouldering »: symptômes B
- mastocytose médullaire isolée (MMI): sans atteinte cutanée

Table 3 B ('Borderline Benign')- and C ('Consider Cytoreduction')-findings useful to assess mast cell burden and aggressiveness of systemic mastocytosis

B-findings[1] Assess disease burden	C-findings[2] Assess disease aggressiveness
1. BM biopsy showing >30% infiltration by MCs (focal, dense aggregates) and/or serum total tryptase level >200 ng/mL	1. BM damage caused by infiltration of neoplastic MCs with consecutive cytopenia(s) (ANC <1.0 × 10^9/L, Hb <100 g/L, or platelets <100 × 10^9/L).
2. Signs of dysplasia or myeloproliferation, in non-MC lineage(s), but insufficient criteria for definitive diagnosis of a hematopoietic neoplasm (AHNMD), with normal or slightly abnormal blood counts.	2. Palpable hepatomegaly with SM-related impairment of liver function, ascites, and/or portal hypertension.
3. Hepatomegaly without impairment of liver function, and/or palpable splenomegaly without hypersplenism, and/or lymphadenopathy by palpation or imaging	3. Skeletal involvement with large (several cm) osteolytic lesions and/or pathological fractures caused by local MC infiltration. 4. Palpable splenomegaly with hypersplenism. 5. Malabsorption with weight loss due to GI MC infiltrates.

Arock et al, Eur J Hematol 2015

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Syndromes myélodysplasiques, syndromes myéloprolifératifs, leucémies aigues, leucémie chronique à éosinophiles (! Rechercher mutation FIP1L1-PDGFRa)
Plus rarement syndrome lympho-prolifératifs

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Sévère, rare, présence de « symptômes C »

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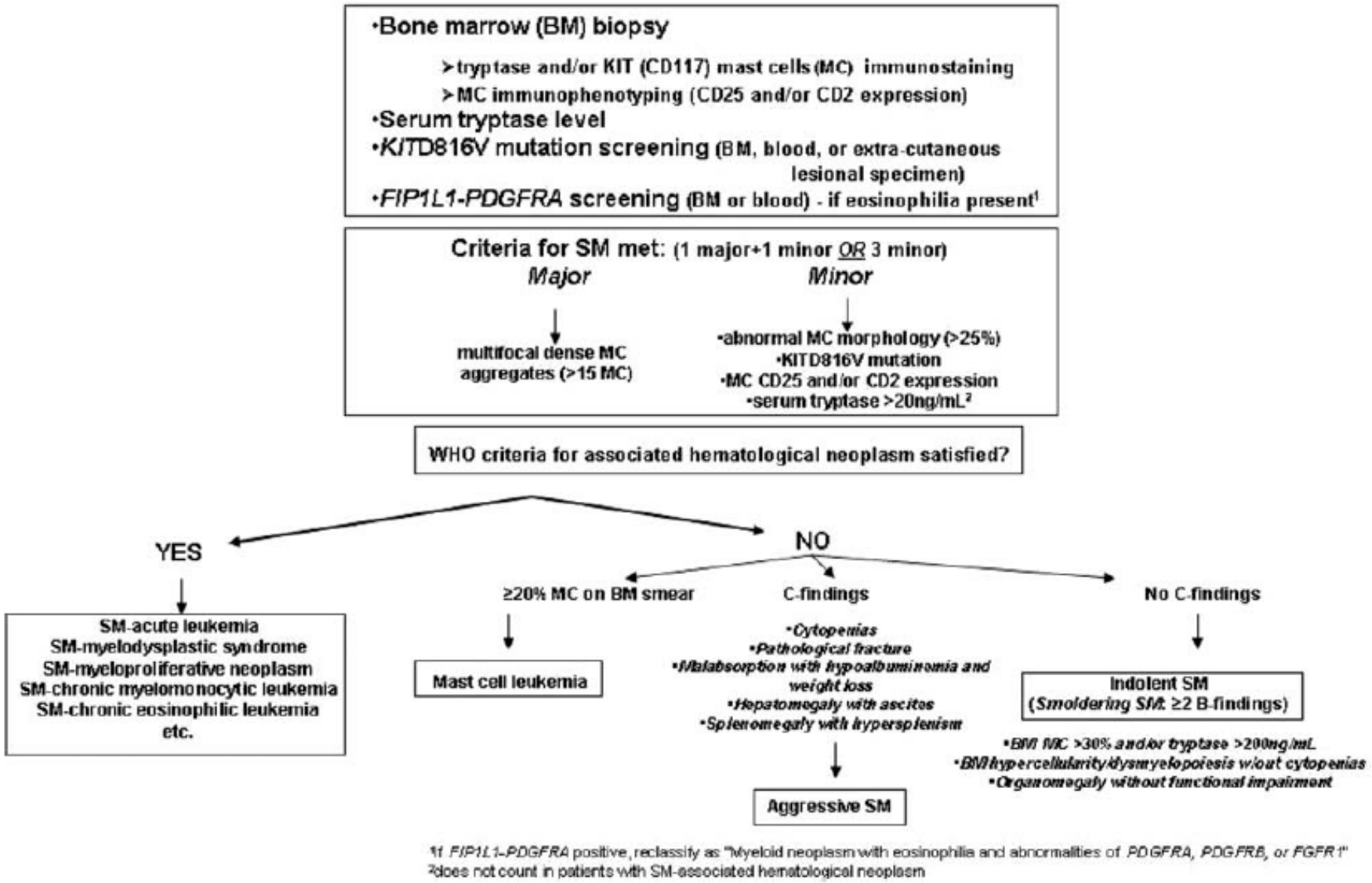
>10% mastocytes sanguins et >20% mastocytes atypiques dans la moelle
Variante « aleucémique »: < 10% dans le sang

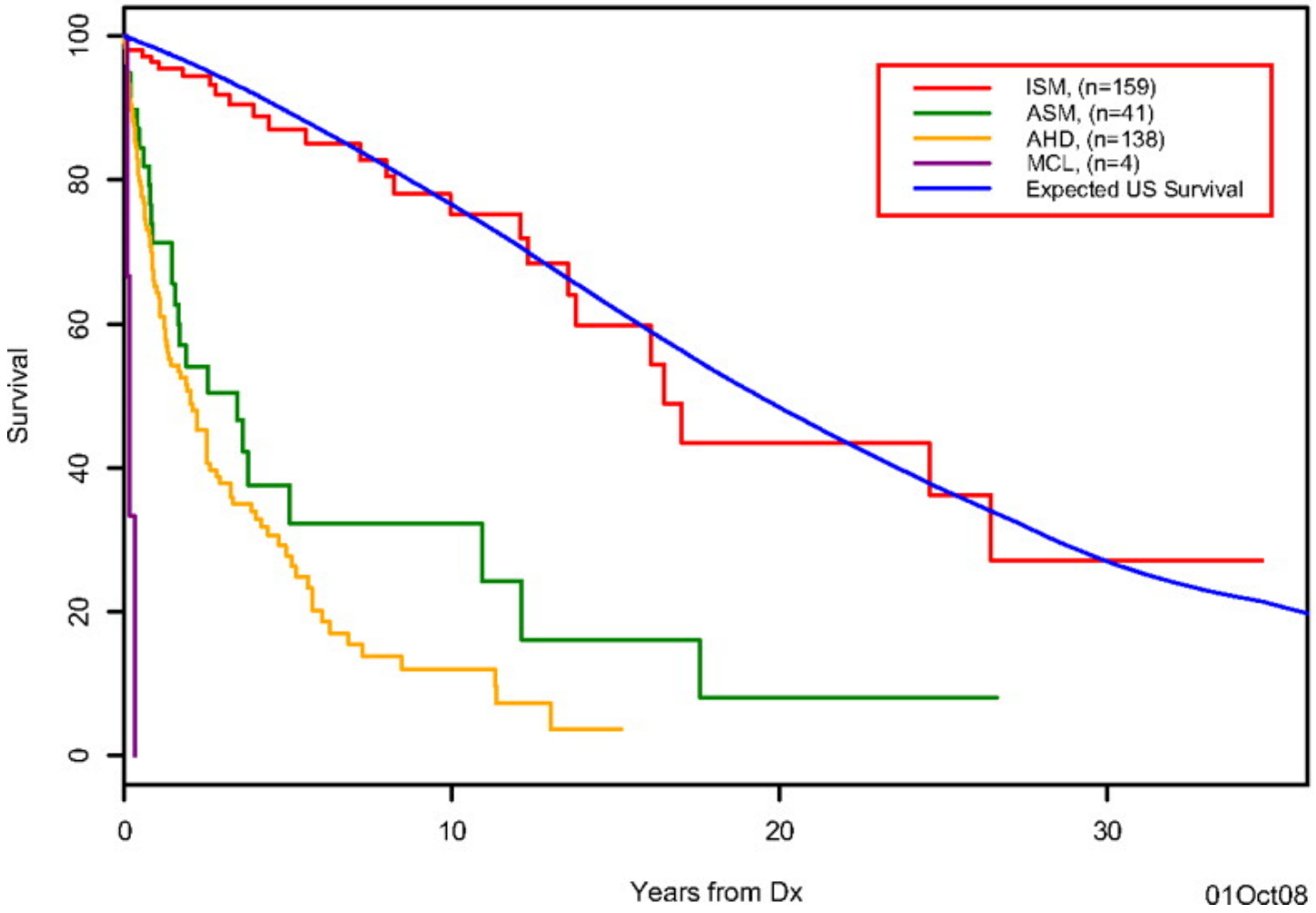
Diagnostic algorithm for systemic mastocytosis (SM)



[1] If *FIP1L1-PDGFRA* positive, reclassify as "Myeloid neoplasm with eosinophilia and abnormalities of *PDGFRA, PDGFRB, or FGFR1*"
[2] does not count in patients with SM-associated hematological neoplasm

Pardanani et al, Am J Hematol 2015

Survie



Lim KH et al, Blood 2009

Traitement

- Au cas par cas!

- Mastocytoses cutanées et systémiques indolentes:
- Contrôler les symptômes d'activation mastocytaire
- Prise en charge multidisciplinaire (hématologue, dermatologue, allergologue, rhumatologue)

- Mastocytoses systémiques plus agressives:
- Traitement cytoréducteurs
- Nouvelles thérapies ciblées (inhibiteurs de tyrosine kinase)

Prise en charge: MC et MSI

1. **Eviter les facteurs déclenchants**

2. Traiter les symptômes systémiques liés à la libération des médiateurs mastocytaires

3. Evaluation et traitement de l'allergie et l'anaphylaxie

Facteurs déclenchants

Encadré 1. Facteurs pouvant favoriser la dégranulation mastocytaire.

Variations thermiques marquées (bains chauds)
Exercice physique intense, traumatismes
Émotions et stress
Venins d'hyménoptères (abeille, guêpe)
Aliments histamino-libérateurs : alcool, œufs, chocolat, fraises, ananas, fruits exotiques, crustacés, poissons, tomates...
Médicaments et apparentés : aspirine (indiquée cependant dans certains cas), anti-inflammatoires non stéroïdiens, anticholinergiques, myorelaxants, opiacés, codéine, codéthyline, procaïne, lidocaïne, polymyxine B, amphotéricine B, quinine, réserpine, hydralazine, pentazocine, thiamine, interféron alpha (indiqué cependant dans certains cas), dextran, mannitol, produits de contraste iodés

Barete S. Ann Derm Vener 2014

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1. Eviter les facteurs déclenchants

2. **Traiter les symptômes systémiques liés à la libération des médiateurs mastocytaires**

3. Evaluation et traitement de l'allergie et l'anaphylaxie

Table 3. Pharmacologic therapies for symptom control in *adult* patients with indolent systemic mastocytosis

Symptom	Treatment ladder*	Drug class	Specific drugs/doses	Common side effects (>5%-10%) and precautions†
Pruritus, flushing	First line	H1-antagonist	Cetirizine, 5-10 mg/d‡ Fexofenadine, 60 mg BID or 180 mg/d‡ Hydroxyzine, 25 mg q 6 h‡ *Doses can be increased with supervision if indicated	Headache, somnolence, confusion, asthenia, xerostomia *Precautions*: Hydroxyzine-anti-cholinergic effects: use with caution in older patients, those with glaucoma, BPH, asthma, and similar conditions
	Second line	Leukotriene antagonist	Montelukast, 10 mg/d Zafirlukast, 20 mg BID	Headache *Precautions*: liver function impairment, neuropsychiatric conditions
	Third line	Nonsteroidal anti-inflammatory drug	Aspirin (see text)	Gastrointestinal bleeding, peptic ulcer disease *Precautions*: may precipitate anaphylactic reaction (see text), aspirin hypersensitivity, children/adolescents with flu (Reye's syndrome), hepatic or renal dysfunction, bleeding disorders
	Third line	Psolaren plus UV A photochemotherapy	See specialized texts	Nausea, pruritus, erythema of varying degree, increased risk of nonmelanoma skin cancers *Contraindications*: Pregnancy, xeroderma pigmentosa, lupus erythematosus with photosensitivity

Pardanani A. Blood 2013

Abdominal pain, cramping, diarrhea, heartburn, nausea, vomiting	First line	H2-antagonist	Ranitidine, 150 mg BID Famotidine, 10 mg BID Cimetidine, 400 mg BID	Headache, abdominal pain, dizziness, constipation, diarrhea *Cimetidine*: gynecomastia
	Second line	Proton pump inhibitor	Omeprazole, 20 mg/d Pantoprazole, 40 mg/d Rabeprazole, 20 mg/d	Headache, abdominal pain, nausea, vomiting, diarrhea, flatulence
	Third line	Sodium cromolyn	100-200 mg QID 30 min before meals and bedtime	Dysgeusia, cough, osmotic diarrhea
	Fourth line	Corticosteroid	Prednisone, 0.5-1 mg/kg/d starting dose; taper as feasible based on response/tolerance	Dose/duration dependent (consult comprehensive drug reference resource)
Headache, cognitive impairment, depression	First line	H1- and H2-antagonist	As above	As above
	Second line	Sodium cromolyn	As above	As above
Recurrent hypotension*	First line	Epinephrine	See text	See text
	Second line	H1- and H2-antagonists	As above	As above
	Third line	Corticosteroid	Prednisone (as above)	As above
	Fourth line	Cytoreductive therapy (IFN-α or 2-chlorodeoxyadenosine)	See text/below	See text/below

Pardanani A. Blood 2013

Osteoporosis	First line	Bisphosphonate	Alendronate, 70 mg q wk	Flu-like symptoms, abdominal pain, nausea, vomiting, diarrhea, asthenia, hypocalcemia, rash, musculoskeletal pain, headache, osteonecrosis of the jaw, nephrotoxicity Follow established guidelines for bisphosphonate use (see text)
			Risedronate, 35 mg q week	*Precautions*: esophageal/upper GI disease (oral bisphosphonates), renal disease, poor oral hygiene or dental procedures
			Pamidronic acid, 90 mg IV q 4 wk Zoledronic acid, 4 mg IV q 4 wk	
	Second line	Cytokine/ immunomodulatory drug	IFN-α, starting dose, 1-3 MU SQ 3 $\times$ per wk; target dose, 3-5 MU SQ 3–5 $\times$ per wk	Dose dependent (consult comprehensive drug reference resource) *Comment*: pegylated interferon may be better tolerated
	Third line	Purine nucleoside analog	2-chlorodeoxyadenosine (cladribine/2-CdA), 5 mg/m^2 IV $\times$ 5 d every 4-8 wk	Myelosuppression, immunosuppression

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Prise en charge: MC et MSI

1. Eviter les facteurs déclenchants

2. Traiter les symptômes systémiques liés à la libération des médiateurs mastocytaires

3. **Evaluation et traitement de l'allergie et l'anaphylaxie**

Allergies et Mastocytose

- Prévalence de l'allergie: = population générale
  (sauf pour allergie aux venins d'hyménoptères)

- Incidence de l'anaphylaxie: > population générale
  20 à 49% selon les études
  Surtout si absence de lésion cutanée et si MSI
  « triggers » : venins d'hyménoptères, nourriture, médicaments (39% idiopathiques)
  Plutôt symptômes d'hypotension que respiratoires

→ EPIPEN® pour tous les patients
→ Omalizumab (Xolair®): si anaphylaxie récurrente
→ Désensibilisation au venin d'hyménoptères: à vie

Bonadonna et al, J Investig Allergol Clin Immunol 2014

Réactions aux venins d'hyménoptères et Mastocytose

Table 3. Prevalence of Clonal Mast Cell Disease in Patients With Systemic Reactions to Hymenoptera Venom Screened on the Basis of Elevated Tryptase

Author	Patients	Elevated Tryptase, No. %	Clonal Mast Cell Disease	%
Haeberli et al 2003[a] [69]	259	19 (7.3)	3 CM	1%
Dubois 2004[b] [47]	2375	32 (1.3)	22 SM	1%
Rueff et al 2006[c] [71]	1102	106 (9.6)	21 CM + 8 SM	2.6%
Bonadonna et al 2009 [24]	379	44 (11.6)	21 ISM + 9 MMAS	7.9%
Potier et al 2009[c] [72]	138	22 (15.9)	1 CM + 5 SM	4.4%
Guevara et al 2010[c,d] [73]	274	30 (10.9)	1 CM + 3 ISM	1.5%

Abbreviations: CM, cutaneous mastocytosis; ISM, indolent systemic mastocytosis; MMAS, monoclonal MC activation syndrome; SM, systemic mastocytosis.
[a]Bone marrow evaluation not performed.
[b]Screening with urinary histamine metabolite.
[c]Evaluation of CD25/CD2 mast cell coexpression and *KIT* mutation not performed or reported.
[d]Bone marrow evaluation performed if serum tryptase >15 ng/mL.

Dosage de tryptase recommandé, mais..
Quel cut-off de tryptase pour lancer le bilan de mastocytose?...

Bonadonna et al, J Investig Allergol Clin Immunol 2014

Table 2. REMA Scoring Model[a]

Variable		Score
Gender	Male Female	+1 –1
Clinical symptoms	Absence of urticaria and angioedema Urticaria and/or angioedema Presyncope and/or syncope	+1 –2 +3
Serum tryptase	<15 ng/mL >25 ng/mL	–1 +2

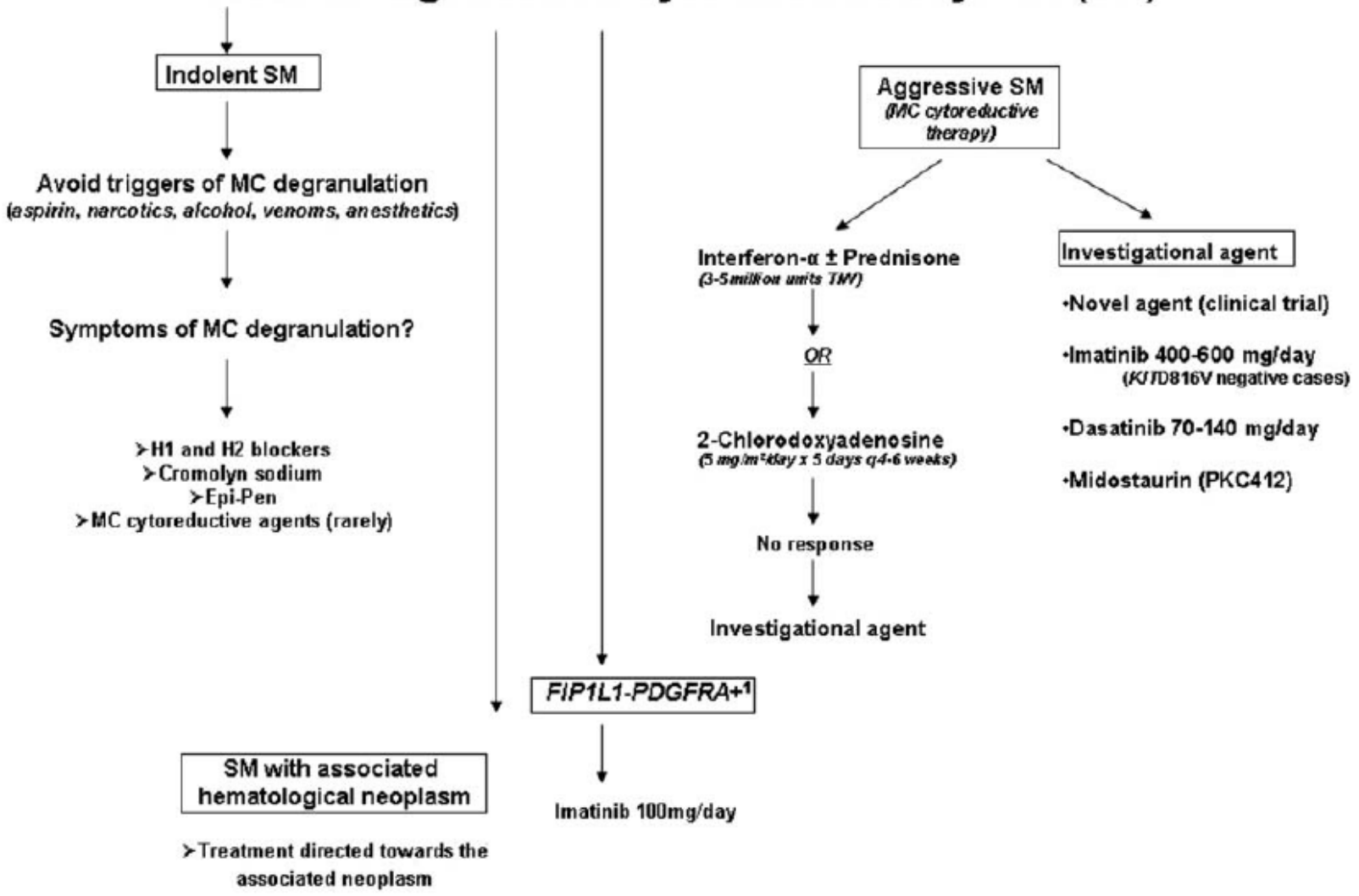
Abbreviation: REMA, Red Española de Mastocitosis (Spanish Network on Mastocytosis).

[a]Proposed as a screening method for the presence of clonal mast cells in patients presenting with anaphylaxis in the absence of cutaneous mastocytosis before a bone marrow study.

Score > ou = 2: bilan complet de mastocytose
Sensibilité 92%
Spécificité 81%

Bonadonna et al, J Investig Allergol Clin Immunol 2014

Treatment algorithm for systemic mastocytosis (SM)



[1]If *FIP1L1-PDGFRA* positive, reclassify as "Myeloid neoplasm with eosinophilia and abnormalities of *PDGFRA, PDGFRB, or FGFR1*"

Pardanani et al, Am J Hematol 2015

Conclusions

- Les mastocytoses constituent un groupe hétérogène de pathologies, dont les manifestations cliniques sont très variées.

- Le diagnostic est difficile, surtout en cas d'absence de lésions cutanées. Il faut y penser en cas d'anaphylaxie inexpliquée et récurrente, de flush, ostéoporose, maladie gastro-intestinale ulcérative ou crampes abdominales récurrentes.

- Le traitement des formes indolentes a pour but d'améliorer la qualité de vie des patients en contrôlant les symptômes d'activation mastocytaire.

- Les nouvelles thérapies ciblant les voies de signalisation du mastocyte, en tenant compte de son statut mutationnel, devraient améliorer dans le futur le pronostic des formes plus agressives.

- Une prise en charge multi-disciplinaire est nécessaire (allergologues, dermatologues, hématologues, rhumatologues), ainsi qu'une information et éducation du patient.

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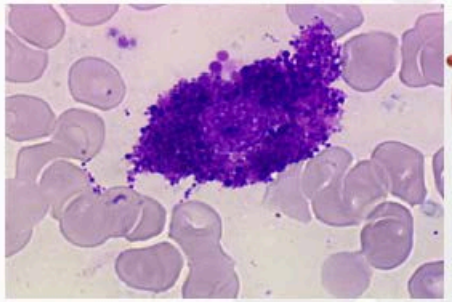

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DESTINE AUX PATIENTS ATTEINTS DE MASTOCYSTOSE ET AUX FAMILLES