Systemic mastocytosis

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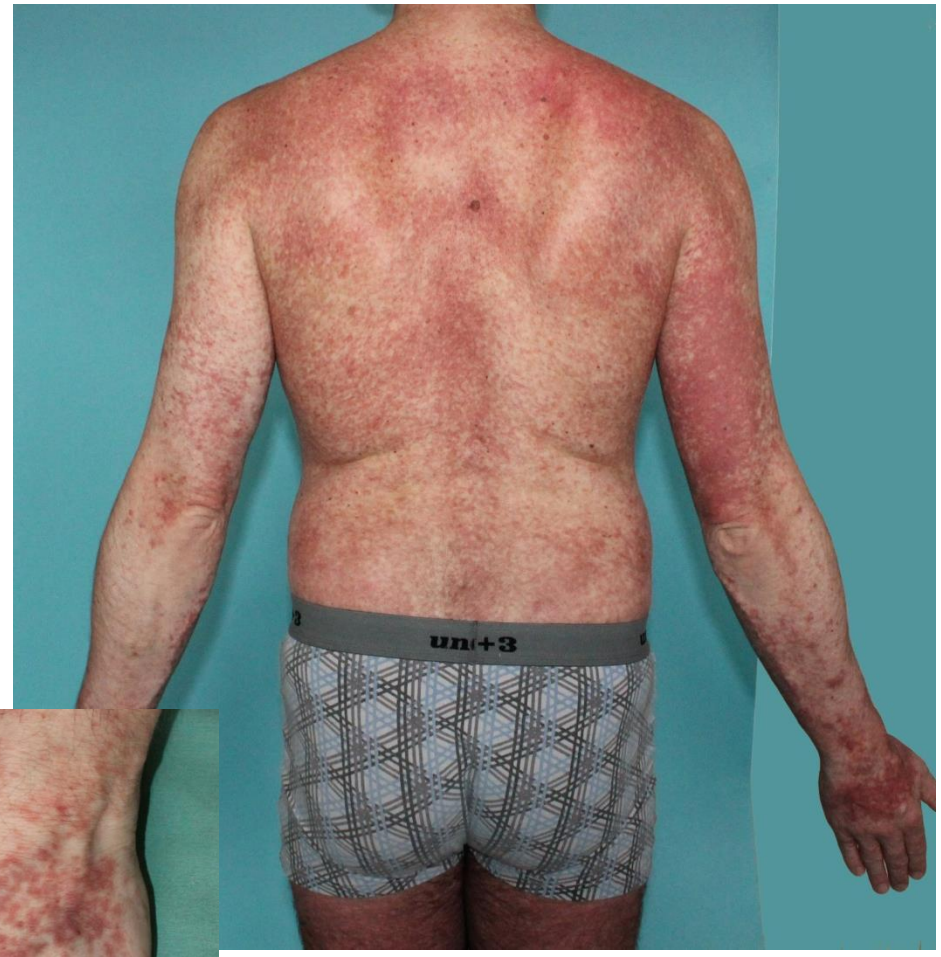
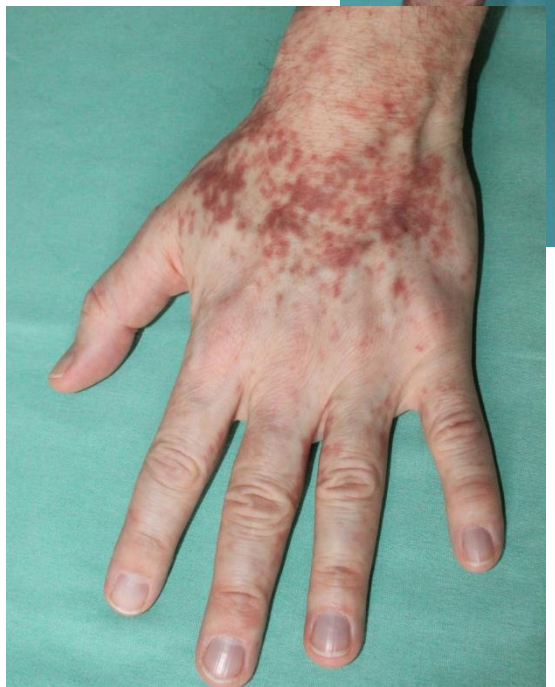
Pacient 49 y. o. with 13 years lasting exanthema

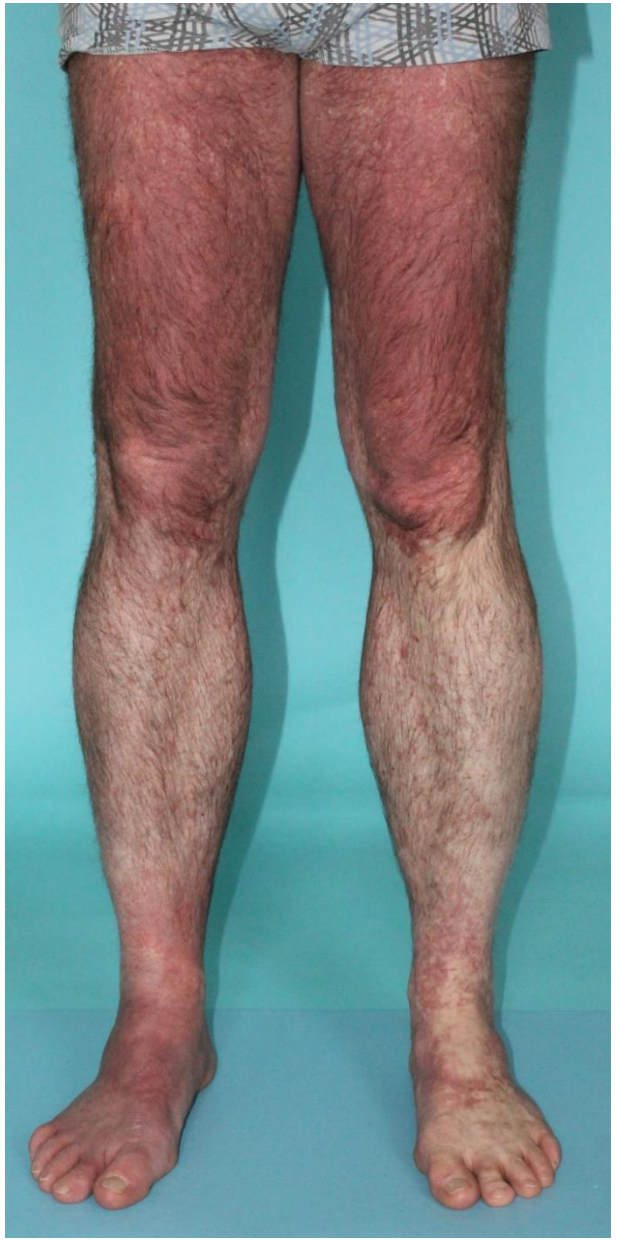
Exanthema is continuous and slowly progressive

Mild itch after warm shower

No GI, cardiac, respiratory, neurologic symptoms, no bone pain

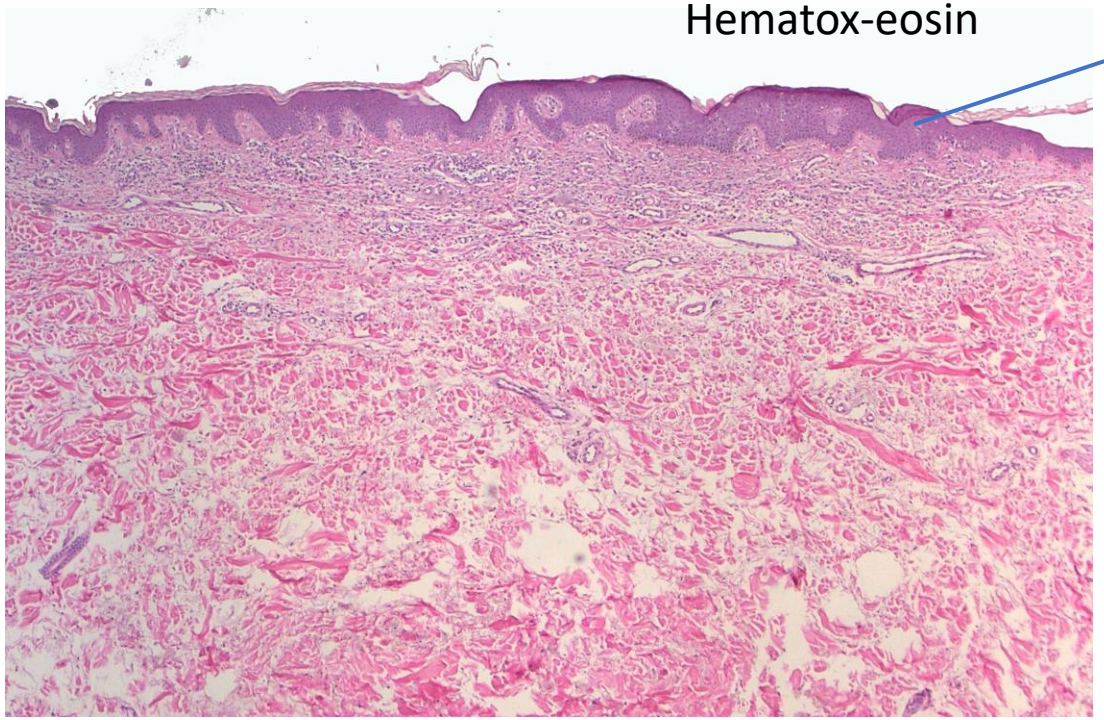




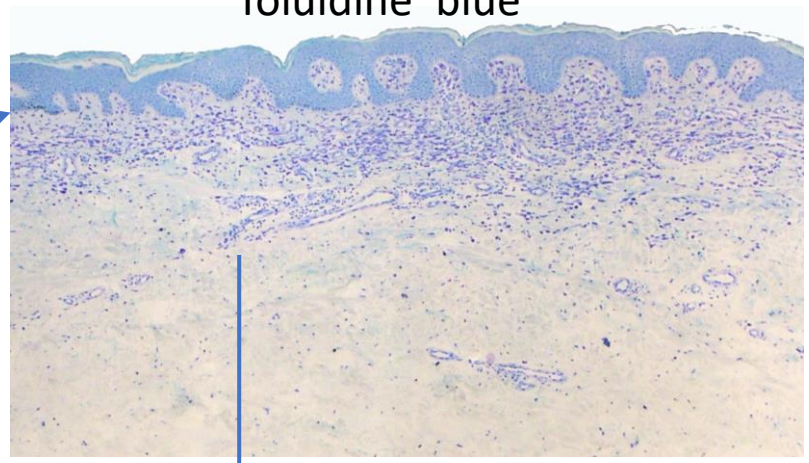


Skin biopsy

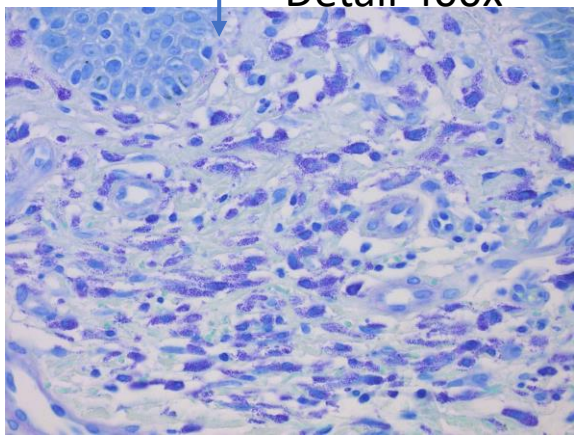
Hematox-eosin



Toluidine blue



Detail 400x



Examination

Skin biopsy

– compatible with urticaria pigmentosa

Sent to hematology to rule out systemic disease

- CT thorax and abdomen – multiple lesions in liver – sonographical not confirmed, activated inguinal lymph nodes – follow–up sonography showed no progression
- Biochemical blood examination normal
- Bone marrow biopsy, c- kit mutation
- **Heterozygous mutation of c-kit, (Asp816Val) confirmed**
- **Bone marrow infiltration with CD 25 positive mastocytes**

- serum tryptase 47,9 mikrog/l

Diagnosis

Fulfilled diagnostic criteria for systemic mastocytosis

indolentn form with bone marrow involvement

Regular follow-up – hematology

Symptomatic therapy (local corticosteroids, antihistamines….)

local symptoms – itch, flush

systemic symptoms – mastocyte mediator release: gastric pain, diarrhea, runny nose, respiratory , cardiovascular – anafylactic reaction, headache , bone pain

References

- Fotoarchive Department of Dermatovenereology First Faculty of Medicine and General University Hospital, Prague
- Bolognia JL., Schaffer JV., Cerroni L.:Dermatology, Elsevier, 4th Edition, 2017
- El Hussein S. et al. Systemic Mastocytosis and Other Entities Involving Mast Cells: A Practical Review and Update, Cancer (Basel). 2022 Jul 17;14(14):3474