Paediatric mastocytosis

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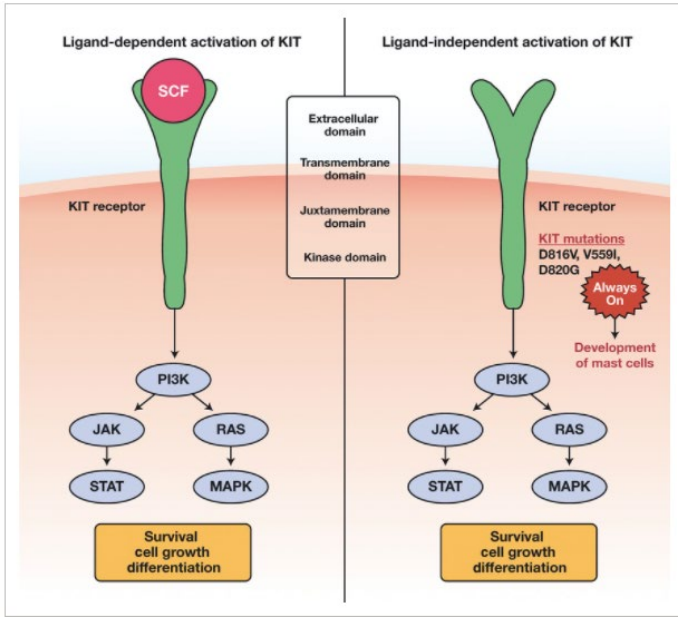
Pathogenesis

- Remains not completely understood
- Somatic activating mutations in the KIT gene are found in >80% of all cases of mastocytosis.
- In about 40 % of children a mutation in exon 17 (codon 816) was found (vs >85% of adults), in about 40% mutations outside exon 17 (exon 8,9,11) were observed, about 20% no KIT-mutation detected
- Mutation in Kit D816 V (Aspartat gets replaced by Valin) -> abnormal accumulation and activation of mast cells
- But:

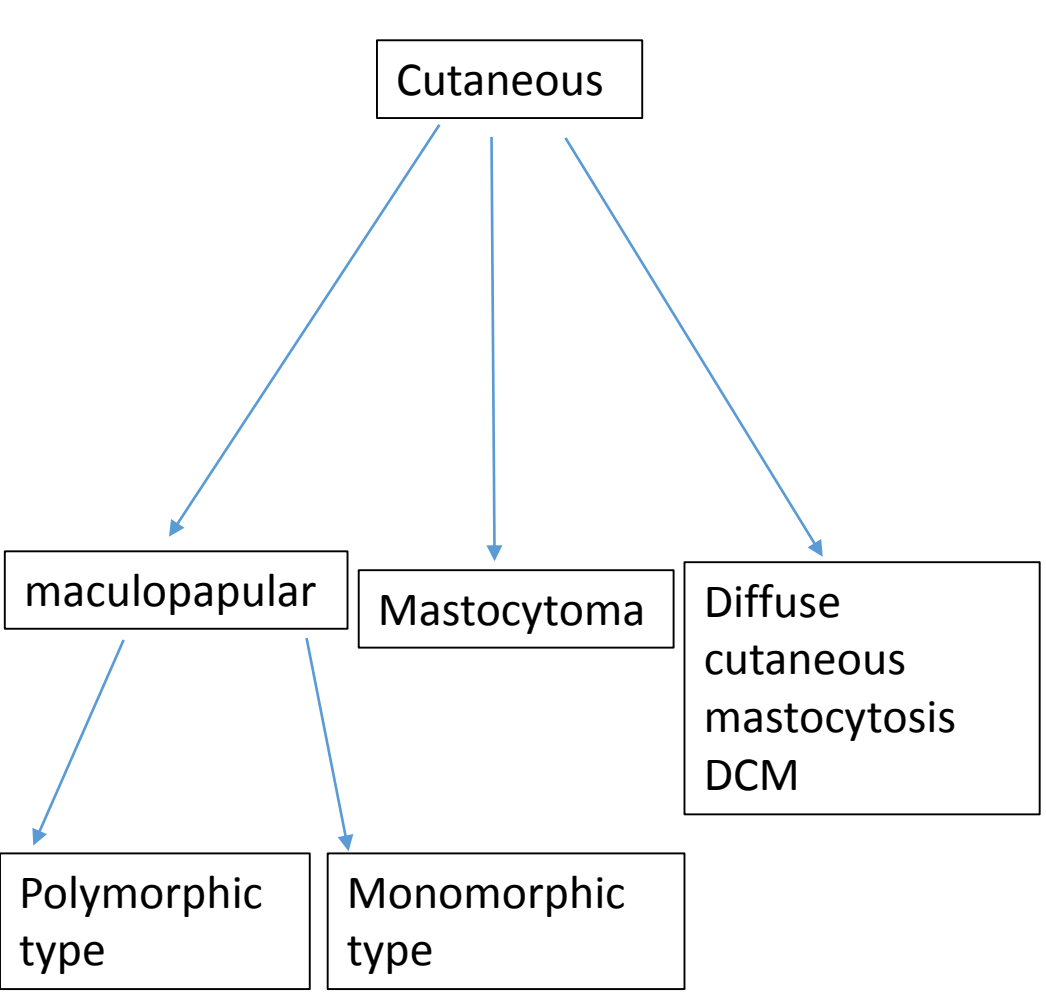
➢ Patients harbouring the same mutation might present with very different clinical pictures (aggressive systemic mastocytosis, indolent SM)

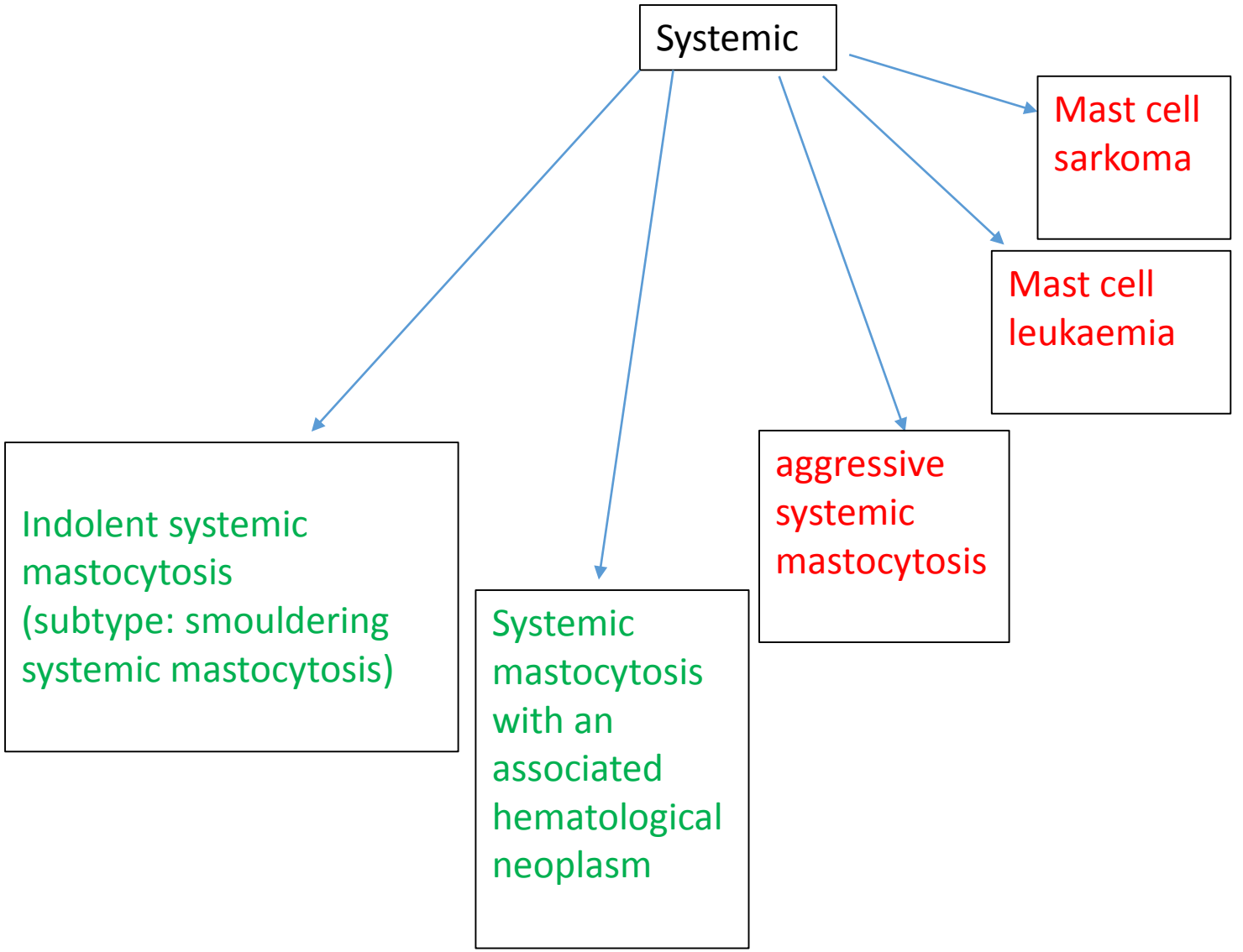
➢ In paediatric mastocytosis, despite the KIT mutation a complete sponatneous remission is generally observed at puberty

- Other factors, other genes?



Classification





Classification cutaneous Mastocytosis

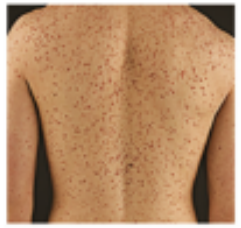
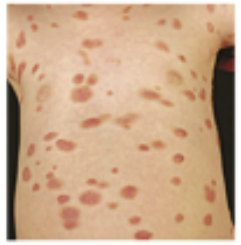
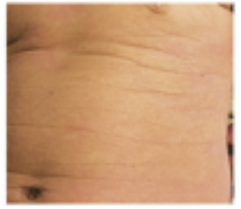
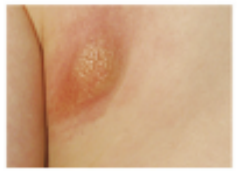
Subforms	Variants	Typical manifestations
Maculopapular cutaneous mastocytosis (syn. urticaria pigmentosa)	Monomorphic	
	Polymorphic	
Diffuse cutaneous mastocytosis		
Cutaneous mastocytoma		

FIG 8. Refined classification of cutaneous involvement in patients with mastocytosis.

Of note:

- Urticaria pigmentosa obsolete
- Teleangiectasia macularis eruptiva perstans no longer used, as not a separate variant
- Term «solitary mastocytoma» no longer used

K.Hartmann et al. J Allergy Clin Immunol 2016

Maculopapular cutaneous mastocytosis (MPCM): **Polymorphic type**

- Majority of paediatric patients

- Brown or red, often oval shaped lesions of different sizes
- Plaques /nodules (term still not optimal)
- Lesions can vary during course of disease (nodules-> plaques -> macules)
- Asymmetric distribution, head, neck and extremities are typically involved
- Brown lesions on lateral part of forehead are characteristic for this form

- Serum tryptase usually not elevated

- Systemic involvement extremely rare

- Prognosis favorable, most patients show spontaneous regression by adolescence

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Maculopapular cutaneous mastocytosis (MPCM): Monomorphic type

- Small percentage of paediatric patients
- Small monomorphic round lesions (like observed in adulthood)
- Possibly increased serum tryptase levels (does not decrease over time)
- Might have systemic involvement
- Usually persists into adulthood

Diffuse cutaneous mastocytosis (DCM)

- Rare
- Large areas of skin are infiltrated by mast cells
- Patients exhibit generalized erythema, usually with pachydermia (thickened skin)
- Extensive blistering and/or exfoliation (DD: bullous dermatosis). Tendancy to blister improves with 3-4 years
- Serum tryptase levels usually increased, but mostly no systemic involvment
- Severe systemic symptoms possible (abdominal pain, hypotension, shock..)
- Lesions resolve/improve by adolescence

Mastocytoma

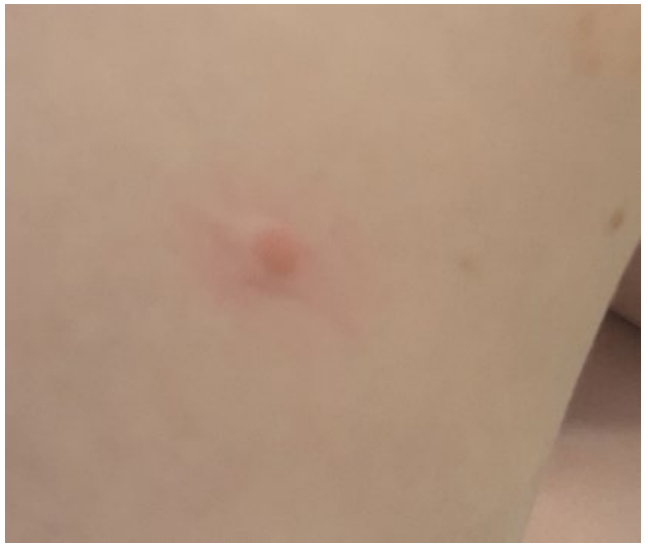
- Most common type of mastocytosis in infants < 3months
- In 40% present at birth
- Solitary or up to three
- Firm, red/yellowish/brown – coloured
- Flare-ups or blisters possible
- Complete regression (almost never in adults)
- DD: juvenile xanthogranuloma, Spitz nevus

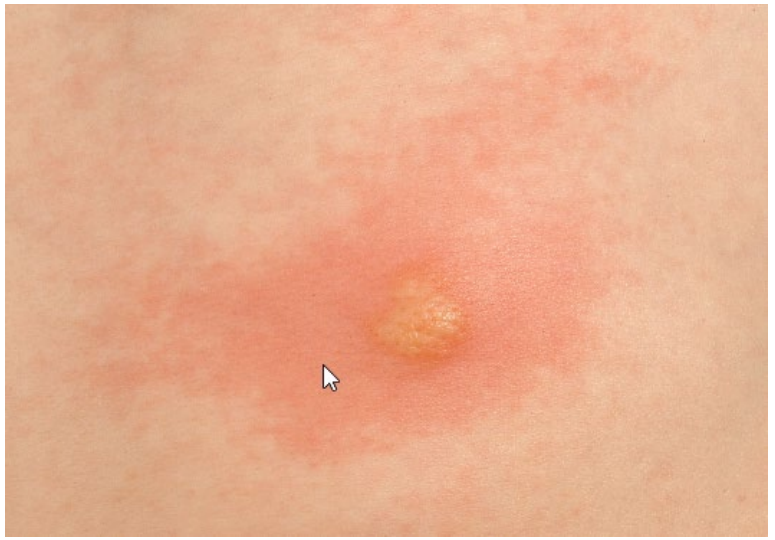
Diagnosis/diagnostic workup

- Typical cutaneous lesions
- Darier-sign: important clinical feature

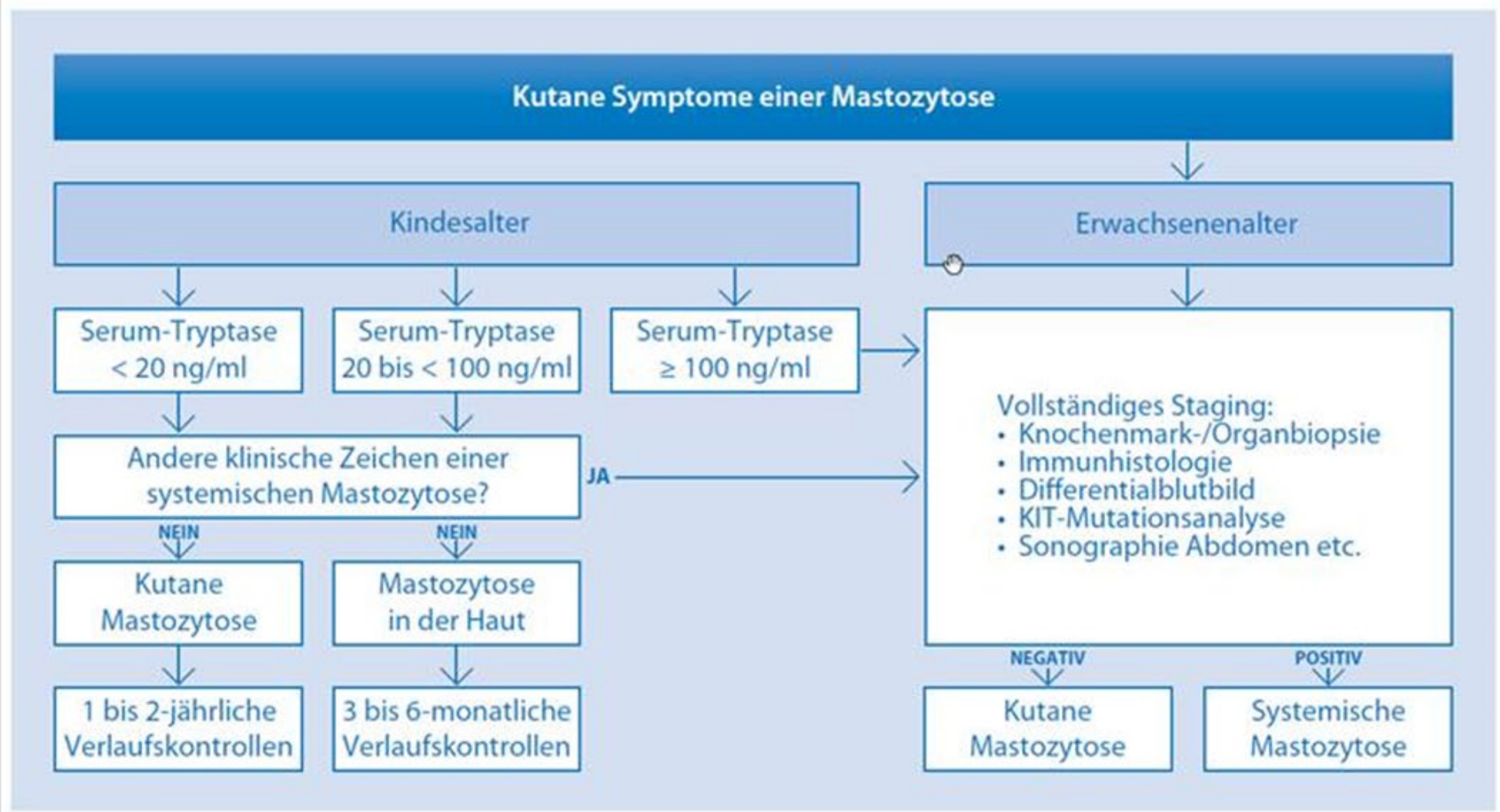
Elicited by stroking a lesion around 5 times by using moderate pressure with a tongue spatula.
Within a few minutes, a wheal-and-flare reaction of the lesion develops

- Serum Tryptase: at time of diagnosis (except: mastocytomas);no recommendations for regular testing
- If unclear: Histology





Tryptase



Ott, Kopp, Lange. Kinderallergologie in Klinik und Praxis. Springer Verlag 2014:

Adulthood-onset and childhood-onset mastocytosis

TABLE I. Characteristics of typical adulthood-onset and typical childhood-onset mastocytosis

Parameter	Adulthood-onset mastocytosis	Childhood-onset mastocytosis
Most frequent category of mastocytosis	ISM	Cutaneous mastocytosis
Typical course of the disease	Chronic	Temporary
Frequency of anaphylaxis (%)	50	<10
Typical tryptase level (μg/L)	>20	<20
Typical location of *KIT* mutation	Exon 17, most frequently *KIT* D816V	Exon 8, 9, 11, or 17 or absent
Most frequent type of cutaneous lesions	Maculopapular	Maculopapular
Typical morphology of maculopapular lesions	Monomorphic	Polymorphic
Typical size of maculopapular lesions	Small	Large
Typical distribution of maculopapular lesions	Thigh, trunk	Trunk, head, extremities

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Emergency kit

Anaphylaxie-Notfallplan

Photo

Name
Vorname
Geburtsdatum

Bekannte Allergene, die eine Anaphylaxie auslösen können:
1.
2.
3.

Im Notfall bitte verständigen:
1.
☎
mobil
2.
☎
mobil

Plan ausgestellt von
Arzt
Datum
Unterschrift

Erste Hilfe bei **beginnender Reaktion**

Anzeichen

- Jucken an Handflächen, Fußsohlen, im Genitalbereich
- Kratzen im Hals
- Schwellung von Lippen, Gesicht
- Hautrötung
- Quaddeln, Nesselausschlag
- Übelkeit, Erbrechen
- Unbestimmtes Angstgefühl

Handlungsmassnahmen

1. Antihistaminikum und Kortison verabreichen
   Name des Antihistaminikums und Menge eintragen:
   Name des Kortisons und Menge eintragen:
2. Den Pen bereit halten
   Notarzt verständigen: 144

Auf weitere Anaphylaxie-Anzeichen hin beobachten

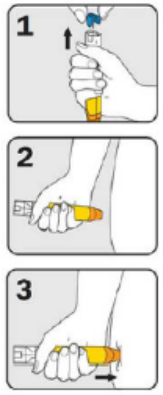
Erste Hilfe bei **schwerer Reaktion**

Anzeichen

- Plötzliche Heiserkeit
- Pfeifende Atmung
- Gleichzeitiges Auftreten von zwei oder mehr Symptomen an verschiedenen Organen (z. B. Bauchkrämpfe und Quaddeln)
- Atemnot
- Bewusstlosigkeit
- Jede Reaktion nach (z. B. Verzehr von Erdnüssen, Wespenstich etc.)

Handlungsmassnahmen

1. Pen in den seitlichen Oberschenkelmuskel injizieren (s. Abb.)
2. Patientenlagerung bei Atemnot: hinsetzen
   bei Kreislaufbeschwerden: hinlegen
   bei Bewusstlosigkeit: stabile Seitenlage
3. Bei Atemnot zusätzlich Spray anwenden - 2 Hübe, ggf. kurzfristig wiederholen
   Name des Sprays eintragen:
4. Notarzt verständigen: 144
5. Zusätzlich Antihistaminikum und Kortison geben (s.o.)



10 Sekunden

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Adaptiert nach:

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INSELGRUPPE

Notfallset Laien

Steroid p.o.

- 0-6a: Betamethason/Betnesol 0.3 mg/kg KG
- > 6a: Prednisolon/Spiricort 2 mg/kg KG

Antihistaminikum p.o. (H1-Antihistaminikum)

- 0-1a: Feniallerg 10 Tropfen
- > 1a: Xyzal Tropfen/Tabletten 1-2a: 10 Tropfen, 2-3a: 15 Tropfen, 3-6a: 20 Tropfen; >6a: 40 Tropfen oder 10 mg

Adrenalin i.m. (alpha-/beta-Agonist)

- Jext® 150 µg (15-30 kg), Jext® 300 µg (>30 kg)
- EpiPen® Junior 150 µg (7.5-25 kg), EpiPen® 300 µg (>25 kg)

DCM, elevated Tryptase, systemic symptoms