



MASTOCITOSI E BPDCN: LE DIAGNOSI DIFFICILI

BOLOGNA, ROYAL HOTEL CARLTON
12 OTTOBRE 2026

Costituzione di un ambulatorio multidisciplinare (mastocitosi)

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Disclosure: Marianna Criscuolo

Company name	Research support	Employee	Consultant	Stockholder	Speakers bureau	Advisory board	Other
Blueprint			X				
GSK			X				
Gilead	X						

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- ❑ A 36-year-old female presents to the clinic for specialist counseling regarding recurrent miscarriages and thrombophilia
- ❑ 3 first-trimester miscarriages, for which she underwent thrombophilia screening that revealed heterozygosity for the prothrombin G20210A mutation and reduced levels of protein S anticoagulant, with subsequent prescription of heparin prophylaxis
- ❑ During the 4th pregnancy, enoxaparin prophylaxis until delivery in March 2022



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- ❑ In May 2022, she was diagnosed with pregnancy-associated osteoporosis following 3 thoracic and 3 lumbar fragility fractures during the postpartum period
- ❑ History of familial hypercalciuria with an episode of nephrolithiasis in adolescence
- ❑ She then underwent a BMD test with T score -3.2. CBC was normal but slight increase of AEC, renal and liver function were normal including serum calcium, vitamin D e PTH

TABLE 3. Definition of Osteoporosis Based on Bone Mass Density¹¹

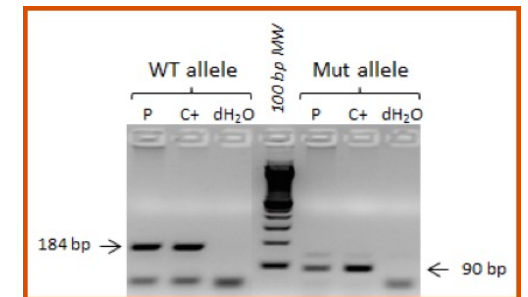
DXA Scores		
Normal	≤1 SD of the mean level for a young-adult reference population	T-score, -1.0 and higher
Low Bone Mass (Osteopenia)	1-2.5 SD below that of the mean level for a young-adult reference population	T-score, -1.0 to -2.5
Osteoporosis	>2.5 SD below that of the mean level for a young-adult reference population	T-score, -2.5 and lower
Severe Osteoporosis	>2.5 SD below that of the mean level for a young-adult reference population	T-score, -2.5 and lower with ≥1 fracture

DXA, dual-energy x-ray absorptiometry; SD, standard deviation

National Osteoporosis Foundation 2020

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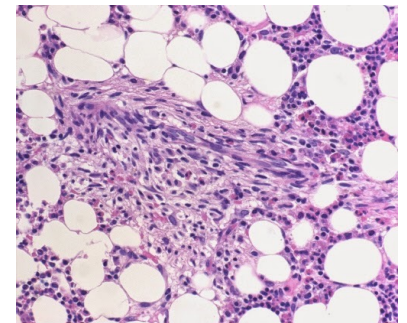
- ❑ At the time of outpatient evaluation she was 11 weeks pregnant w/o heparin prophylaxis
- ❑ Serum tryptase: 28 ng/ml confirmed in subsequent determination
- ❑ PB KIT D816V mutation: positive; FIPL1/PDGFRa rearrangement negative
- ❑ Indication to perform bone marrow biopsy, after delivery



Ematologia^{IN} Progress

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- ❑ Allergology consultation for preferred and avoided medications in analgesia
- ❑ Multidisciplinary management of pregnancy: term delivery, w/o complications
- ❑ Six months after delivery, she underwent a bone marrow biopsy, which confirmed the suspected diagnosis of systemic mastocytosis
- ❑ Referred for specialist evaluation for osteoporosis follow-up



Bonadonna et al ENDA/EAACI position paper Allergy 2015

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Multidisciplinary medical team (MDT):

A group of healthcare professionals from various disciplines involved in comprehensive care
Each brings their expertise to contribute to treatment decision according to their individual role



HEMATOLOGY
ADVANCEMENTS

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STEP 1

Team's interpersonal relationships

STEP 2

Collaborative approach to address all aspects of a patient's condition

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STEP 3

Regular meetings and clear communication to inform all team members

STEP 4

Institutional recognition of MDT as a medical activity

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ADVANTAGES

- ☐ knowledge sharing
- ☐ interactions with colleagues
- ☐ reviewing the patient file
- ☐ helping with decision-making
- ☐ patient satisfaction

DISADVANTAGES

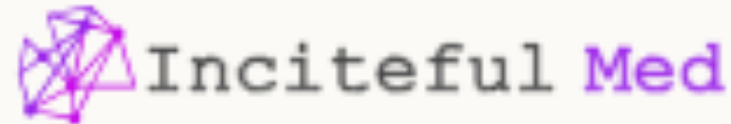
- ☐ technical issues
- ☐ exacerbation of problems within teams
- ☐ peer judgments
- ☐ no specific time for this activity
- ☐ time consuming

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The Essential Specialists

A well-rounded care team for SM typically includes the following specialists, each focusing on a different aspect of the disease:

- **Hematologist/Oncologist:** Usually the “lead” doctor, especially for advanced forms of the disease. They focus on the bone marrow, monitor your **KIT mutation** levels, and oversee systemic treatments like targeted therapies (TKIs) [3][4].
- **Allergist/Immunologist:** Focuses on the “mediator” symptoms caused by chemical release. They are experts in managing anaphylaxis, chronic itching, and flushing, and they prescribe medications like antihistamines and omalizumab [4][6].
- **Dermatologist:** Given that skin symptoms (like urticaria pigmentosa) are often the most visible and uncomfortable part of SM, a dermatologist plays a crucial role. They help monitor skin lesions, distinguish between skin-only disease and systemic involvement, and offer specialized therapies for skin itching and cosmetic concerns [7][8].
- **Gastroenterologist:** Essential if you experience chronic diarrhea, abdominal pain, malabsorption, or weight loss. They manage the heavy burden of “secretory diarrhea” and may perform endoscopies or colonoscopies to check if mast cells have infiltrated the lining of your digestive tract [9][10].
- **Hematopathologist:** A behind-the-scenes expert who specializes in blood and bone marrow. They are the ones who look at your biopsy samples to confirm the diagnosis using the **WHO 2022 criteria** [3][11].



Questions to Ask Your Doctor

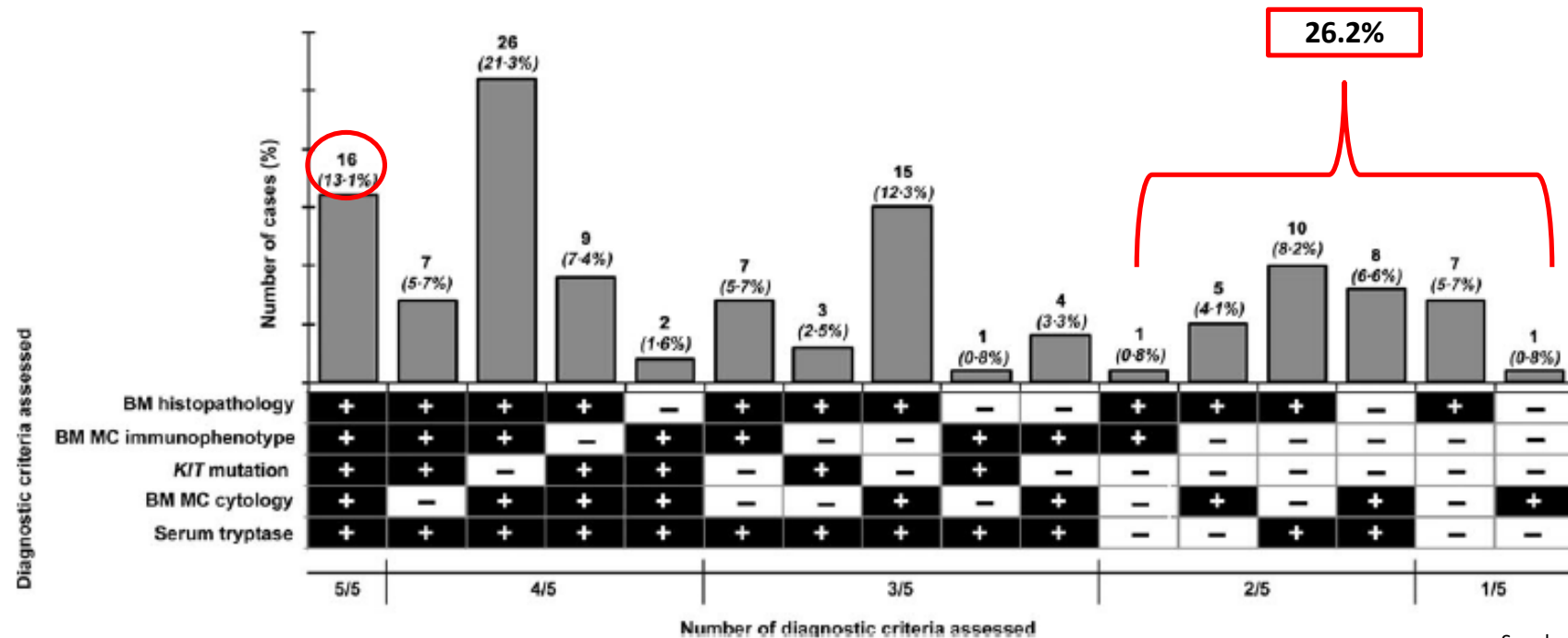
Curated prompts to bring to your next appointment.

1. Which of my doctors is the primary 'quarterback' for my care, and how do you all communicate with each other?
2. Are you familiar with the WHO 2022 diagnostic criteria and how it changes the classification of my specific subtype?
3. What is your experience in managing patients on targeted therapies like avapritinib or midostaurin?
4. How do you determine if a symptom should be treated by an allergist (mediator-related) or a hematologist (disease-burden related)?
5. Do you have a relationship with a Mastocytosis Center of Excellence for second opinions or clinical trial access?

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bjh research paper

Diagnosis and classification of mastocytosis in non-specialized versus reference centres: a Spanish Network on Mastocytosis (REMA) study on 122 patients



Sanchez-Munoz L BJH 2016

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	WHO major criterion	WHO minor criteria				Number of criteria evaluated		
	Immunohistochemical	Morphological	Immunophenotypical	<i>KIT</i> D816V mutation	Serum tryptase >20 ng/ml	5/5 criteria studied	4/5 criteria studied	≤3/5 criteria studied
Referring centre								
Total cases investigated	106 (87%)	86 (70%)	64 (52%)	38 (31%)*	108 (89%)	16 (13%)	44 (36%)	62 (51%)
Non-evaluable	2 (1.8%)	3 (3.4%)	4 (6.2%)	0	0			
Positive	68 (65%)	49 (59%)	40 (67%)	11 (29%)	78 (72%)			
Negative	36 (35%)	34 (41%)	20 (33%)	27 (71%)	30 (28%)			
REMA centre								
Total cases investigated	122 (100%)	122 (100%)	122 (100%)	119 (97.5%)+	122 (100%)	119 (98%)	3 (2%)	0
Non-evaluable	3 (2.5%)	4 (3.3%)	1 (0.8%)	3 (2.5%)	0			
Positive	89 (73%)	102 (84%)	105 (86%)	104 (87%)	89 (73%)			
Negative	30 (25%)	16 (13%)	16 (13%)	15 (13%)	33 (27%)			
p-value ~ (investigated versus non investigated cases)	0.00002	<0.00001	<0.00001	<0.00001	0.00008	<0.00001	<0.00001	<0.00001
p-value ~ (positive versus negative cases)	0.15	0.00002	0.0002	<0.00001	0.90			

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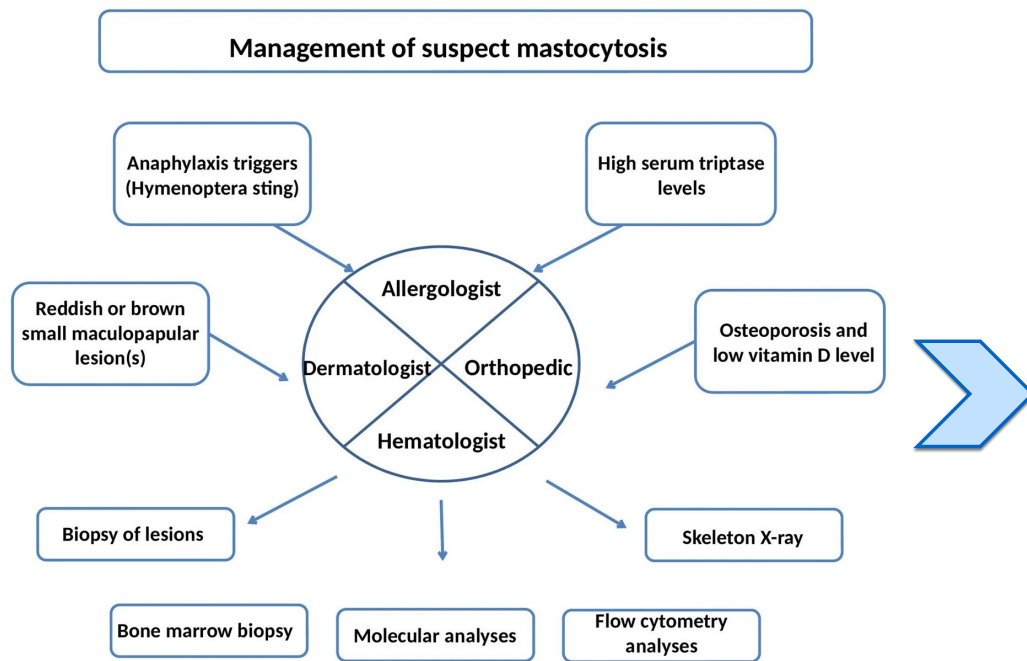
Criterion	Overall agreement % (95% CI)	P-value (McNemar chi-square test)	Kappa coefficient	Overall agreement score
Immunohistochemical	80% (72–89%)	0.01	0.51	moderate
Morphological	68% (56–80%)	<0.0001	0.23	low
Immunophenotypical	75% (62–87%)	0.007	0.31	low
<i>KIT</i> D816V+ mutation	34% (8–60%)	<0.0001	0.04	very low
Serum tryptase >20 ng/ml	90% (84–96%)	1.00	0.74	good

Sanchez-Munoz L BJH 2016

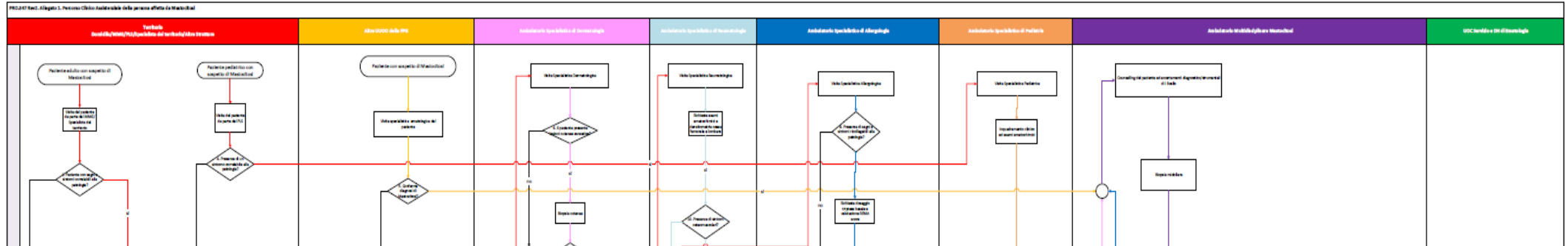
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Management of suspect mastocytosis



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- ☐ **Disease related needs:** dermatology, allergology, hematology and hemopath
- ☐ **Patients request:** pediatricians, ob-gyn
- ☐ **Symptoms control:** gastroenterology, rheumatology, cardiology

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MISSION

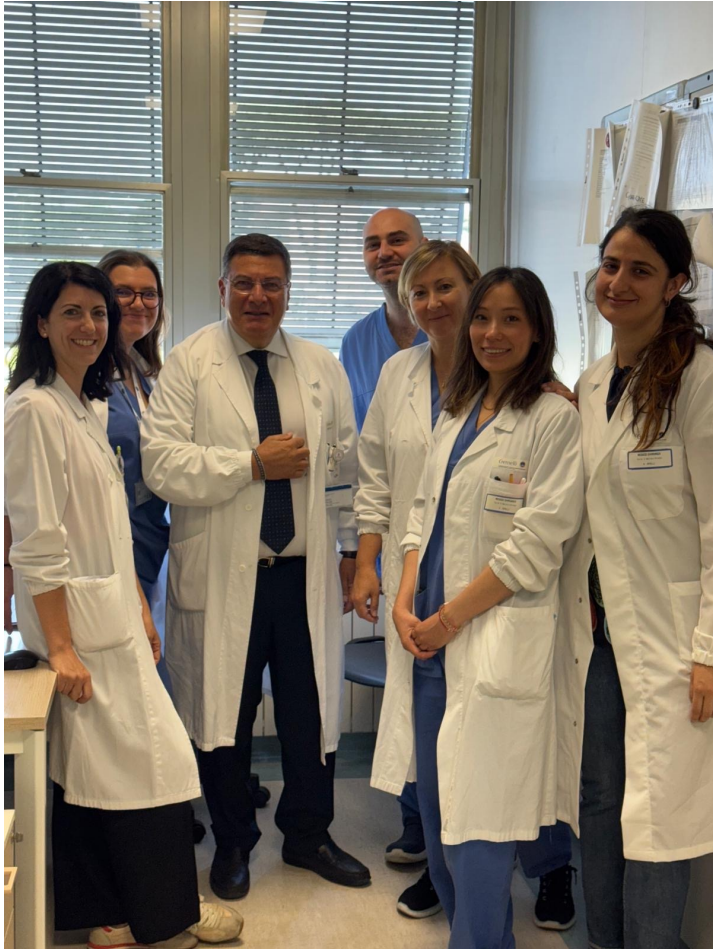
L'associazione Rete Italiana Mastocitosi in acronimo "Associazione RIMA" si è costituita come associazione ai sensi degli articoli 36 e seguenti del Codice Civile in ottobre 2019 con lo scopo di:

- favorire lo sviluppo, l'approfondimento e la divulgazione delle discipline che hanno come oggetto lo studio della mastocitosi
- riunire tutti gli specialisti che si occupano di mastocitosi, coinvolgendo specialisti di vario ambito, in particolare allergologi, ematologi, immunologi, reumatologi, dermatologi, pediatri, gastroenterologi, ginecologi, anestesisti, anatomopatologi, biologi molecolari, etc.
- collaborare strettamente con l'Associazione dei pazienti affetti da mastocitosi (ASIMAS)
- mettere in comunicazione i centri di maggiore e lunga esperienza con i centri che seguono un minor numero di pazienti o da un periodo più breve
- diffondere un modello di cura di tipo multidisciplinare che permetta un miglior trattamento della malattia e di tutte le complicanze ad essa correlata



- ☐ Find the gap: geographical disparity, clinical unmet needs
- ☐ MDT and/or PDTA within the hospital
- ☐ Regional network to discuss single patient charts and improve specific setting
- ☐ Scheduled clinical updates as scientific meeting and annual conference

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Prof. Livio Pagano

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Dr. Antonio Giordano

Dr.ssa Martina Quattrone

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Dr.ssa Angela Rizzi

Prof.ssa Clara De Simone

Dr.ssa Silvia Bosello

Dr. Consolato Gullì

Dr. Gianluca Ianaro

Dr.ssa Mariannita Gelsomino

Patients and their patience

Thank you for your attention

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