FONDAZIONE
ITALIANA
SINDROMI
MIELODISPLASTICHE



CONVEGNO FISiM

Firenze, CSF Montedomini
“Il Fuligno”
23-24 ottobre 2026

FONDAZIONE ITALIANA SINDROMI MIELODISPLASTICHE



Vexas-map

Gabriele Todisco

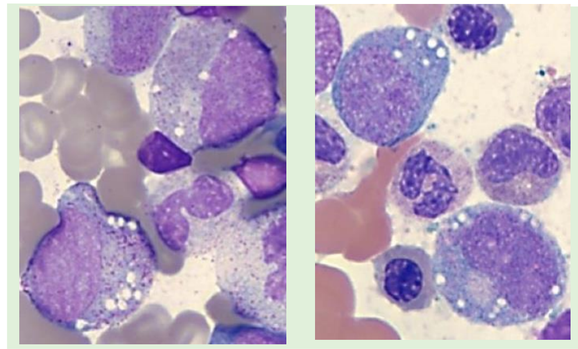
U.O. Oncologia medica ed Ematologia, Humanitas Cancer Center, Istituto Clinico Humanitas IRCCS, Rozzano (MI)

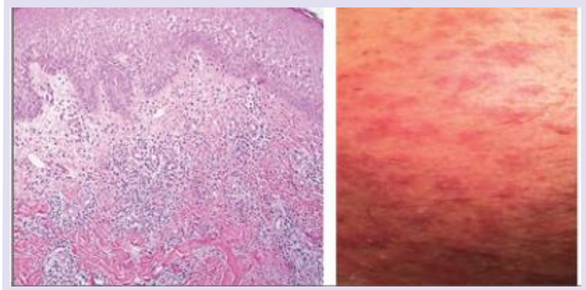
Disclosures of Gabriele Todisco

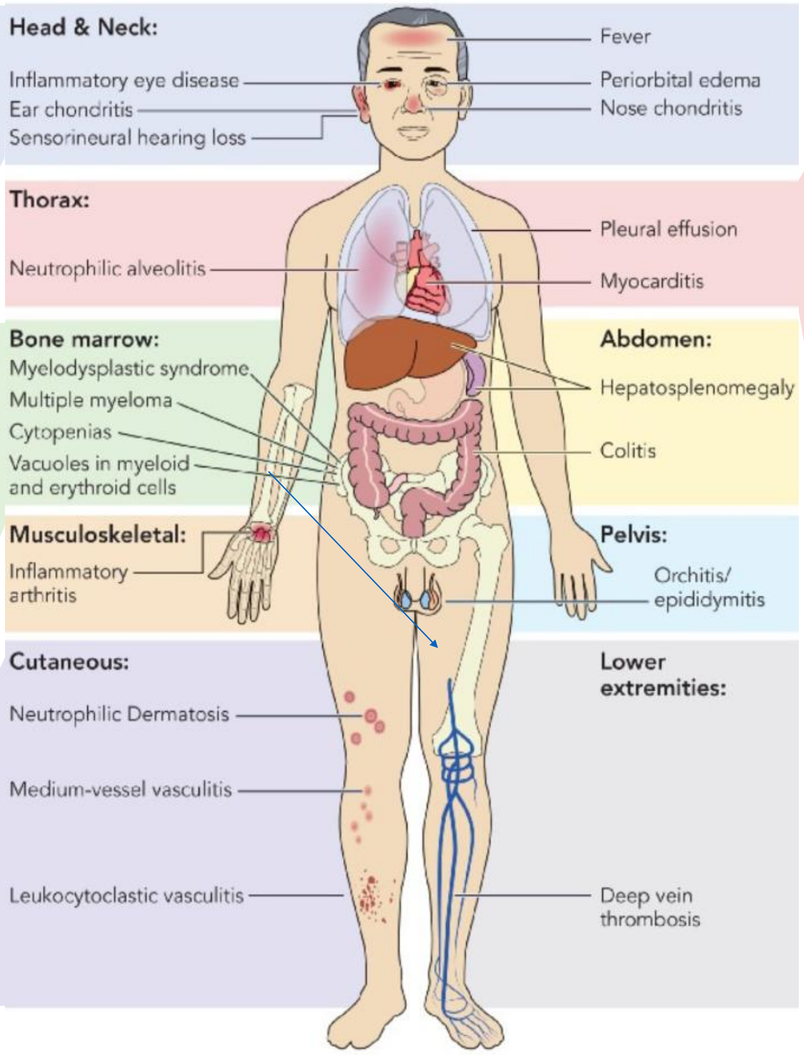
Company name	Research support	Employee	Consultant	Stockholder	Speakers bureau	Advisory board	Other

VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome

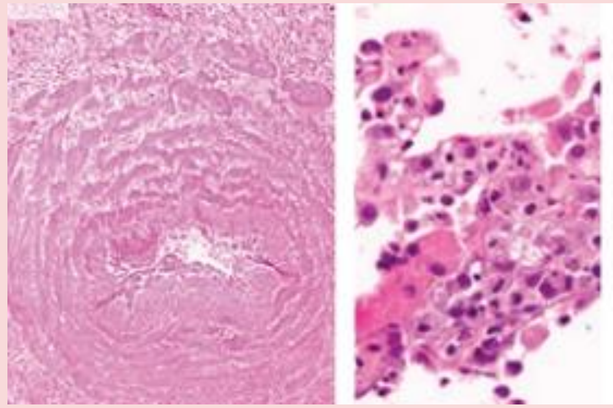












Beck D et al, N Engl J Med 2020;383:2628-38.

VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome

PRESENTAZIONE CLINICA

Segni	
Anemia (macrocitica)	95%
Febbre ricorrente	82%
Dermatosi neutrofila	76%
Artrite sieronegativa	60%
Trombocitopenia	50%
Calo ponderale	50%
Policondrite	40%
Adenopatie mediastiniche	35%
Alveolite polmonare	37%
Trombosi venosa profonda	36%
Vasculite leucocitoclastica	30%

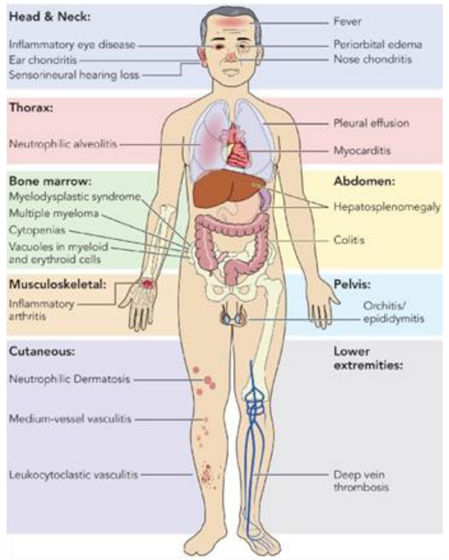
Epidemiologia	
Prevalenza	1/5000 uomini (>50anni)
	1/25000 donne (>50anni)
Età media	74 anni
Maschi	99%

Neoplasie ematologiche associate	
MDS	50%
MGUS	10-20%
MBL	10%

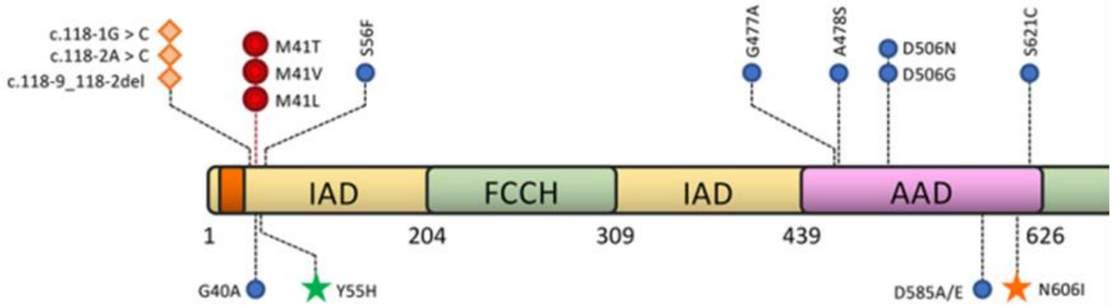
Ann. Rheum Dis 2024;83:1358-1367
Am J Clin Pathol 2024;162:28-40

VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome

DIAGNOSI



+

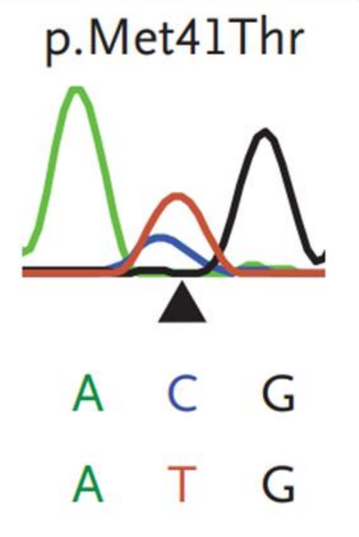


Sanger sequencing

sensibilità

VAF > 10-20%

hotspot



NGS (targeted DNA-seq)

sensibilità

VAF > 1%

25 esoni



VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome

TERAPIA

- **Target 1: *treat the clone***
  - Azacitidina
  - Allo-SCT

- **Target 2: *treat the inflammatory process***
  - Steroide
  - MTX
  - Anti IL1 (anakinra)
  - Anti IL6 (tocilizumab)
  - Anti JAK2 (ruxolitinib, baricitinib)

FONDAZIONE ITALIANA SINDROMI MIELODISPLASTICHE

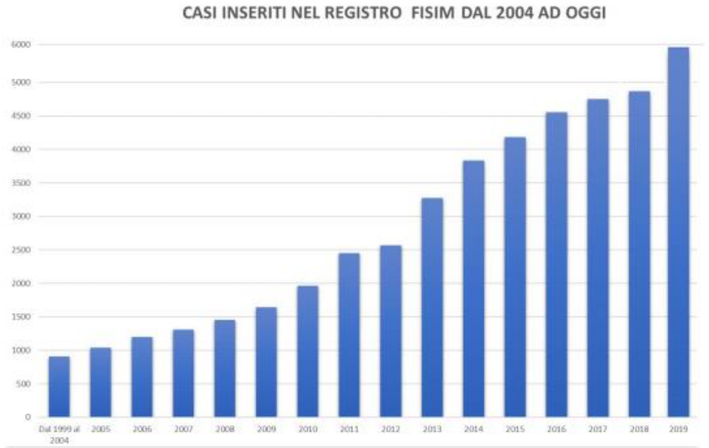
HOME CHI SIAMO MDS e TERAPIE CENTRI REGISTRO RICERCA CONGRESSI E EVENTI NOTIZIE COSA PUOI FARE CONTA'



Il registro FISiM è un progetto iniziato come Gruppo Piemontese per lo Studio delle Sindromi Mielodisplastiche (MDS Piemonte), nato su base volontaristica con intenti scientifici nel 1999. Vi hanno aderito fino al 2010 esclusivamente tutte le divisioni ospedaliere ed universitarie di Ematologia del Piemonte e la maggior parte delle divisioni di Medicina Interna della Regione interessate alla gestione di malati ematologici. Dal 2011 oltre ai centri operanti in Piemonte, si è costituita una rete italiana del registro MDS che comprende tutte le regioni italiane che al momento ne hanno richiesto l'adesione (Basilicata, Calabria, Campania, Emilia-Romagna, Friuli Venezia Giulia, Lazio, Liguria, Lombardia, Marche, Piemonte, Puglia, Sardegna, Sicilia, Toscana) e promosso dalla Fondazione.

Questo progetto consiste nella raccolta di casi con una nuova diagnosi di sindrome mielodisplastica effettuata presso le istituzioni partecipanti e registrati prospetticamente nei singoli registri regionali, tramite l'utilizzo della piattaforma informatica REDCap.

Per ogni paziente che dà il proprio consenso si inseriscono informazioni riguardanti i dati clinici e di laboratorio. I dati in questione sono quelli relativi alla Storia clinica del



ps://www.fisimematologia.it/registro-fisim/

Data Collection Instrument	Arruolamento 12-11-2018	VEXAS	+ Add new Followup 05-06-2019 Vivo	L1 Protocollo sperimentale
Anagrafica	●			
Anamnesi e Esame obiettivo	●			
Diagnosi Outcome E Prognosi	●		●	
Diagnosi - Esame Sangue Periferico	●		●	
Diagnosi - Esame Midollo	●		○	
Diagnosi Score	●		○	
Suspicion of VEXAS	○			
VEXAS		○		
Analisi NGS				
Terapia				●
Cicli ESA				○
Cicli Azacitidina				○
Cicli Lenalidomide				○
Cicli Luspatercept				○
Trasfusioni durante terapia Luspatercept				○
Adverse Event Page				



Registro MDS v.1.1 PID 14

Actions: Download PDF of instrument(s) Video: Basic data entry

Suspicion of VEXAS

Editing existing Codice Paziente **34-1.** 00450001, 1940, F

Event: **Arruolamento** 12-11-2018

Codice Paziente	34-1
Does the patient have a suspicion of VEXAS?	(H) (M) ◉ Yes ○ No reset
Form Status	
Complete?	(H) Complete

Save & Exit Form | Save & ... | – Cancel –

Data Collection Instrument	Arruolamento 12-11-2018	VEXAS	+ Add new Followup 05-06-2019 Vivo	L1 Protocollo sperimentale
Anagrafica	●			
Anamnesi e Esame obiettivo	●			
Diagnosi Outcome E Prognosi	●		●	
Diagnosi - Esame Sangue Periferico	●		●	
Diagnosi - Esame Midollo	●		○	
Diagnosi Score	●		○	
Suspicion of VEXAS	●			
VEXAS		○		
Analisi NGS				
Terapia				●
Cicli ESA				○
Cicli Azacitidina				○
Cicli Lenalidomide				○
Cicli Luspatercept				○
Trasfusioni durante terapia Luspatercept				○
Adverse Event Page				

VEXAS

Editing existing Codice Paziente **34-1**. , ,

Event: **VEXAS**

Codice Paziente	34-1
Record ID	

2. Initial Diagnosis

2.1 Symptom Onset

Date of first symptoms	Today D-M-Y
First symptom (free text)	
Date of VEXAS diagnosis date	Today D-M-Y

2.2 First Specialist Contact

First specialty consulted	○ General medicine ○ Rheumatology ○ Hematology/Oncology ○ Dermatology ○ Other reset

2.3 Clinical Manifestations at Diagnosis

General Symptoms

	Present
Fatigue	☐
Fever	☐
Unintentional weight loss	☐
Night Sweats	☐

Skin

	Present
Neutrophilic dermatosis / Sweet syndrome	☐
Cutaneous vasculitis	☐
Erythema nodosum	☐
Angioedema	☐
Exanthema	☐
Other skin involvement	☐

Lymphatic System		
		Present
Lymph node involvement	Ⓗ 💬 Ⓜ	☐
Cervical	Ⓗ 💬 Ⓜ	☐
Axillary	Ⓗ 💬 Ⓜ	☐
Mediastinal	Ⓗ 💬 Ⓜ	☐
Retroperitoneal	Ⓗ 💬 Ⓜ	☐
Spleen		
		Present
Splenomegaly	Ⓗ 💬 Ⓜ	☐

Lung		
		Present
Infiltrates	Ⓗ 💬 Ⓜ	☐
Pleural effusion	Ⓗ 💬 Ⓜ	☐
Chronic cough (>8 weeks)	Ⓗ 💬 Ⓜ	☐
Other lung involvement	Ⓗ 💬 Ⓜ	☐

Musculoskeletal

	Present
Arthralgia	☐
Polychondritis	☐
Nose	☐
Ear	☐
Epiglottis	☐
Ribs / chest wall	☐
Synovitis	☐
Myalgia	☐
Arthritis	☐
Other musculoskeletal	☐

Destructive Chondritis

	Present
Destructive Chondritis	☐

Eye		
		Present
Uveitis	Ⓗ 💬 Ⓜ	☐
Scleritis	Ⓗ 💬 Ⓜ	☐
Episcleritis	Ⓗ 💬 Ⓜ	☐
Other ocular	Ⓗ 💬 Ⓜ	☐
Heart		
		Present
Pericarditis	Ⓗ 💬 Ⓜ	☐
Myocarditis	Ⓗ 💬 Ⓜ	☐
Other cardiac	Ⓗ 💬 Ⓜ	☐

Vascular		
		Present
Vasculitis	Ⓗ 💬 Ⓜ	☐
Aortitis	Ⓗ 💬 Ⓜ	☐
Aneurysm	Ⓗ 💬 Ⓜ	☐
Other vascular	Ⓗ 💬 Ⓜ	☐

Thromboembolic Events		
		Present
Pulmonary embolism	H M	☐
Deep vein thrombosis	H M	☐
Superficial thrombosis	H M	☐
Arterial thrombosis	H M	☐
Other thrombosis	H M	☐

Gastrointestinal		
		Present
Abdominal pain	Ⓗ 💬 Ⓜ	☐
Chronic diarrhea	Ⓗ 💬 Ⓜ	☐
Ascites	Ⓗ 💬 Ⓜ	☐
Ulceration	Ⓗ 💬 Ⓜ	☐
Peritonitis	Ⓗ 💬 Ⓜ	☐
Other GI	Ⓗ 💬 Ⓜ	☐

Hematologic Manifestations

	Present
Myelodysplastic syndrome	☐
Multiple myeloma	☐
MGUS	☐
CML	☐
Anemia	☐
Macrocytosis	☐
Thrombocytopenia	☐
Neutropenia	☐
Lymphopenia	☐
CCUS	☐
Other hematologic	☐

Transfusion Need

	Present
Anemia requiring transfusion	☐

3. UBA1 Mutation Analysis (Baseline)

3.1 Mutation Testing

Date of UBA1 analysis	Today D-M-Y
Sample type	☐ Blood ☐ Bone marrow ☐ Other
UBA1 mutation present	○ Yes ○ No reset

3.2 Mutation Details

Protein mutation	☐ Met41Leu ☐ Met41Val ☐ Met41Thr ☐ Other
cDNA mutation e.g.UBA1 p.M41V, c.121A>G	
Variant allele frequency (%)	

4. Therapies (Non-hematologic)

4.1 Supportive Care

RBC transfusions units in the last three months	
Platelet transfusions units in the last three months	
Other supportive therapy	

4.2 Immunosuppressants

	Present
Methotrexate	☐
Azathioprine	☐
Mycophenolate	☐
Cyclophosphamide	☐
Other	☐

4.3 Biologics

	Present
IL-1 inhibitors	☐
IL-6 inhibitors	☐
Other biologics	☐

4.4 Steroids

Steroids (in Prednisone-equivalent)		
Treatment Start	**Start date:** Today D-M-Y	**Start dose:**
	Dose unit:	**Frequency:**
Treatment End	**Therapy status:**	**End date:**
	Reason end:	**Specify other:**
Dose Changes	**Changes in dose:** No 1 2 3 reset	
Dose change (1)	**Start date:**	**Start dose:**
	Dose unit:	**Frequency:**
	Reason:	**Specify other:**

Dose Changes	**Changes in dose:** ○ No ○ 1 ○ 2 ○ 3 reset	
Dose change (1)	**Start date:**	**Start dose:**
	Dose unit:	**Frequency:**
	Reason:	**Specify other:**
Dose change (2)	**Start date:**	**Start dose:**
	Dose unit:	**Frequency:**
	Reason:	**Specify other:**
Dose change (3)	**Start date:**	**Start dose:**
	Dose unit:	**Frequency:**
	Reason:	**Specify other:**
Best Clinical Response - **Major**: complete resolution of all clinical symptoms - **Minor**: partial resolution of clinical symptoms	**Response:**	**Date of evaluation:** Today D-M-Y

5. Therapies (Hematologic)

5.1 JAK2 Inhibitors - Drug Selection

	Present
Tofacitinib	☐
Baricitinib	☐
Ruxolitinib	☐
Upadacitinib	☐
Fedratinib	☐
Abrocitinib	☐
Pacritinib	☐
Momelotinib	☐
Other	☐

5.2 Hypomethyleting Agents - Drug Selection

	Present
Azacytidine (s.c.)	☐
Decitabine (i.v.)	☐
Other	☐

5.3 Other - Specify Drug and administrative route

Specify Drug	
Administrative route	

Other

Treatment Start	**Start date:** Today D-M-Y	**Start dose:**
	Dose unit:	**Frequency:**
Treatment End	**Therapy status:**	**End date:**
	Reason end:	**Specify other:**
Dose Changes	**Changes in dose:** No 1 2 3 reset	

Clinical Response - **Major**: complete resolution of all clinical symptoms - **Minor**: partial resolution of clinical symptoms	**Response:**	**Date of evaluation:** Today D-M-Y
Overall Hematological Response	**Hemoglobin response:**	**Neutrophil response:**
	Platelet response:	**Transfusion independence:**
	Date of evaluation: Today D-M-Y	
MRD / Molecular Response		**Performed:** Yes No reset

Form Status

Complete? Incomplete

Save & Exit Form | Save & ...

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