VEXAS syndrom
(D46.7)

Øyvind Midtvedt
Revmatolog
OUS

Læringsmål

Økt kunnskap om VEXAS syndrom

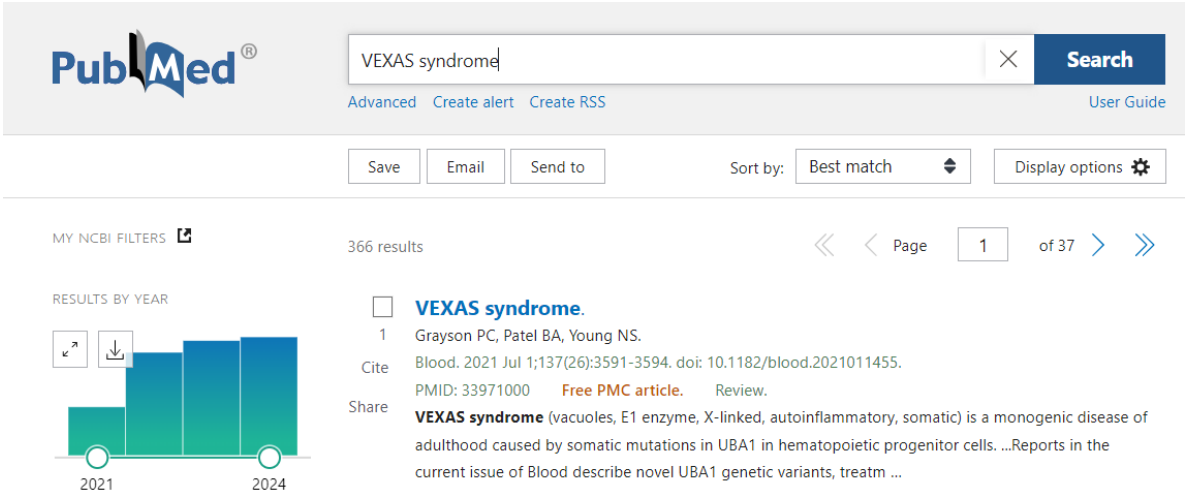
- Symptomer
- Diagnostikk
- Behandling

VEXAS har sett deg, men du har ikke sett VEXAS

Foto pinterest.com

Kunnskapsgrunnlag

PubMed:

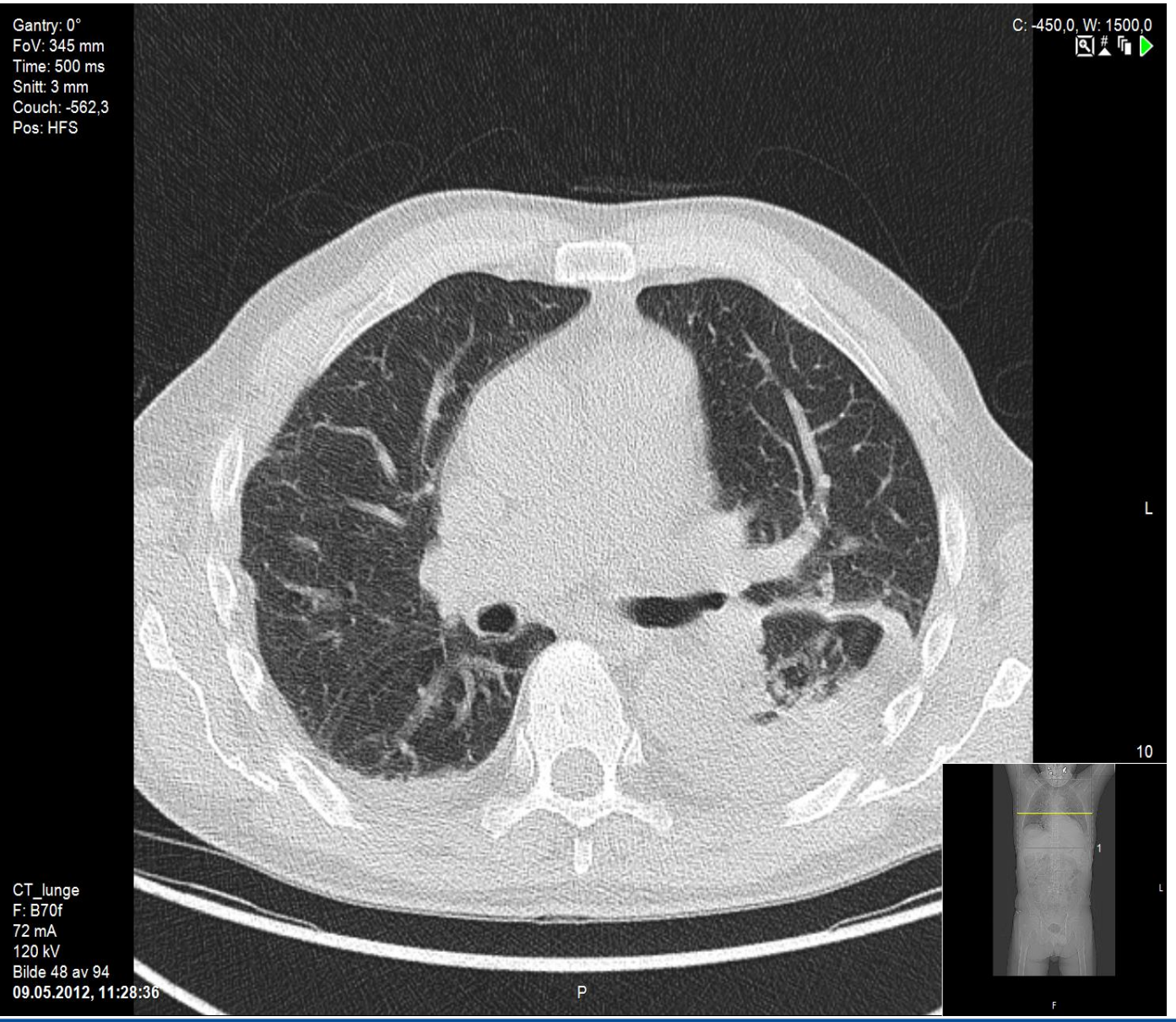


Erfaring fra OUS

Foto Ø. Midtvedt

«Per» 68 år

- Innlagt revmatologisk seksjon OUS 09.05.12
- Tidligere sykdom: Hypertensjon
- Siste 6 mndr redusert allmenntilstand, vekttap, forbigående feber, iridocyklitt
- Brystsmerter, negativ koronar angiografi
- SR 131, CRP 227
- Fortetning ve underlapp, antibiotika behandlet uten effekt
- Temporalisbiopsi x 2 (negativ)
- CT veggfortykkelse av a.subclavia, storkarsvaskulitt?



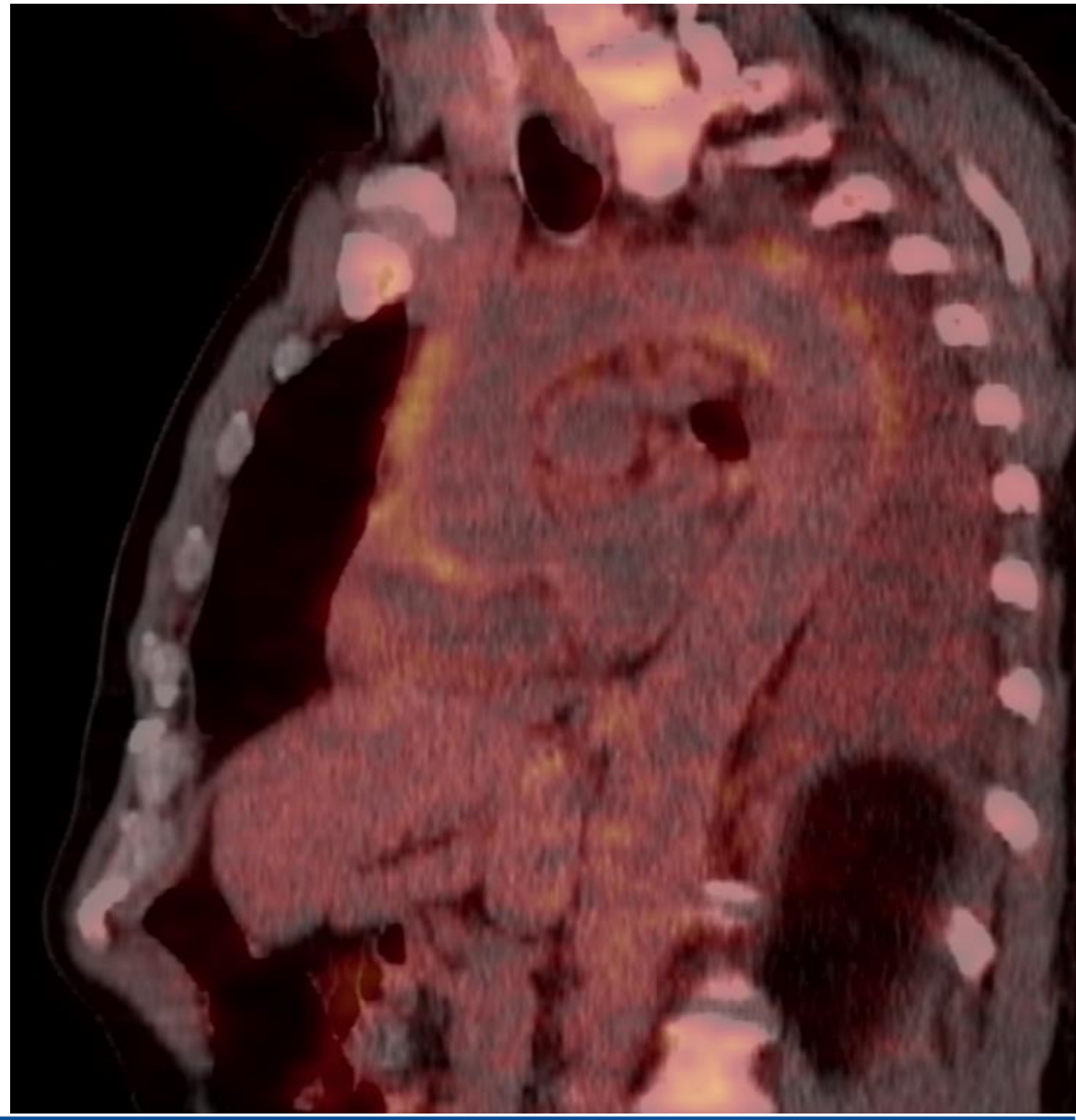


Foto med tillatelse fra ektefelle

Prøvesvar og diagnose

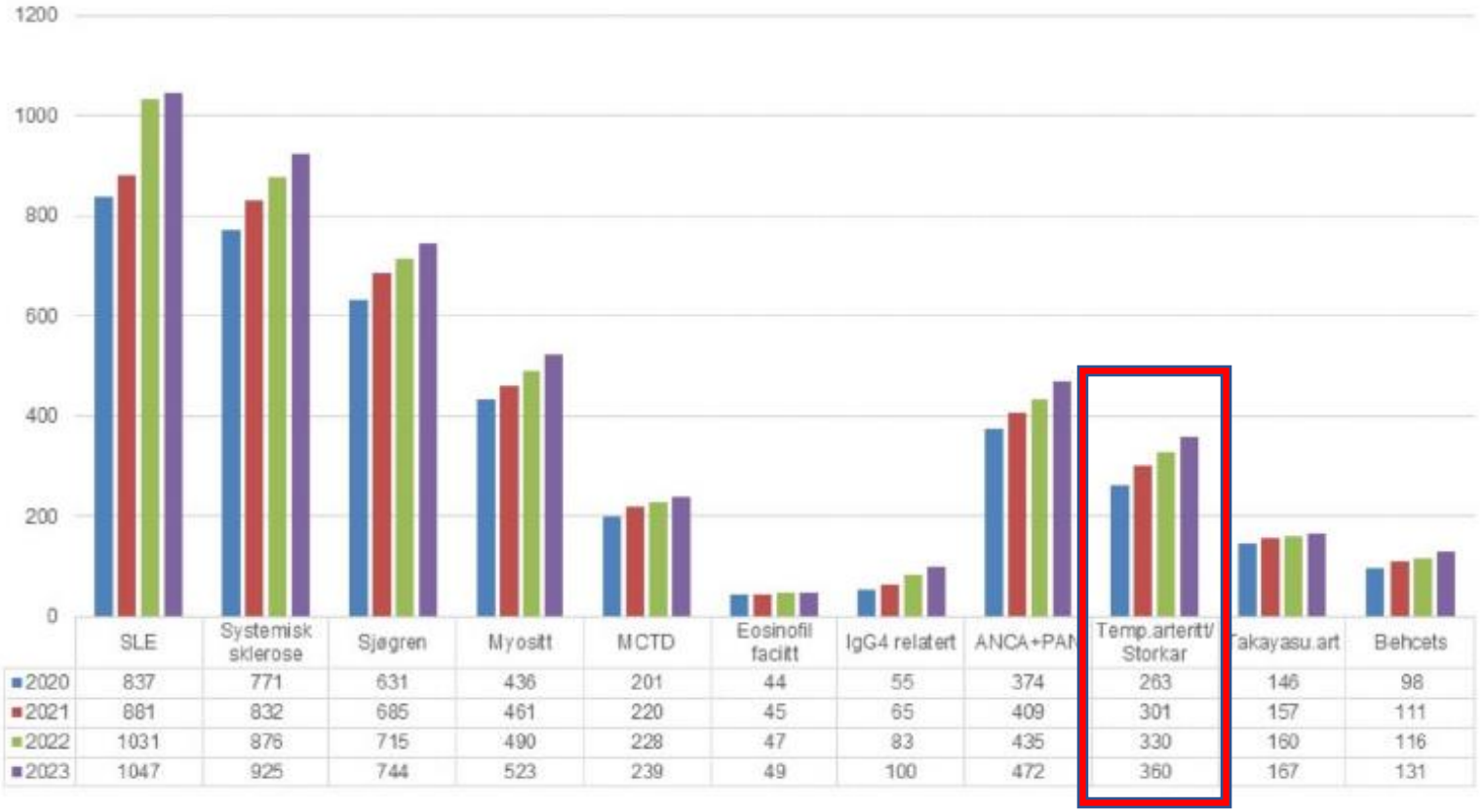
- **Lab:** SR >100,CRP 183
- Hb 9,9, MCV 96 (82-98), trombocytter 496 (145-390), leukocytter 7,8 (3,5-10)
- IgG4 3,0, negativ ANCA, ANA, APS, ingen monoklonal gammopati, neg u-stix

- **Diagnose**: Non-kraniell kjempecellearteritt (KCA, temporalis arteritt)
- KCA insidens 16,7 pr 100 000 > 50 år[1]
- Litt atypisk med iridocyklitt, lungefortetning, forhøyet IgG4
- Prednisolon 1mg/kg fra 09.05.12

1.Brekke et al Arthritis Res Ther.2017

NOSVAR (Norsk Systemisk bindevevssykdom og Vaskulitt Register)

Figur 2: Figuren viser det årlige økende antall pasienter for et utvalg av diagnoser i NOSVAR.



	SLE	Systemisk sklerose	Sjøgren	Myositt	MCTD	Eosinofil facilitt	IgG4 relatert	ANCA+PAN	Temp.arteritt/ Storkar	Takayasu.art	Behcets
2020	837	771	631	436	201	44	55	374	263	146	98
2021	881	832	685	461	220	45	65	409	301	157	111
2022	1031	876	715	490	228	47	83	435	330	160	116
2023	1047	925	744	523	239	49	100	472	360	167	131

2012	2013	2014	2015	2016	2017	2018	2019	2020

Klinikk:

Iridocyklitt — Kondritt øre — Sweets — Mors

Redusert allmenntilstand — Diagnose: **Relapserende Polykondritt**

Feber

Brystsmerter

Biokjemi:

SR 131,CRP 183,

Hb 9,9 — 7,0 — Blodtransfusjon hver 14/d — hver 7-10/d

MCV 96 ,fra sept-12 MCV >100 — (EPO, G-CSF)

Trombocytter 496 — 60 — Revolade

Behandling:

Prednisolon 60mg/d – minste dose 17,5mg/d →

Methotrexate

RoActemra

Kineret - utslett

Infliksimab

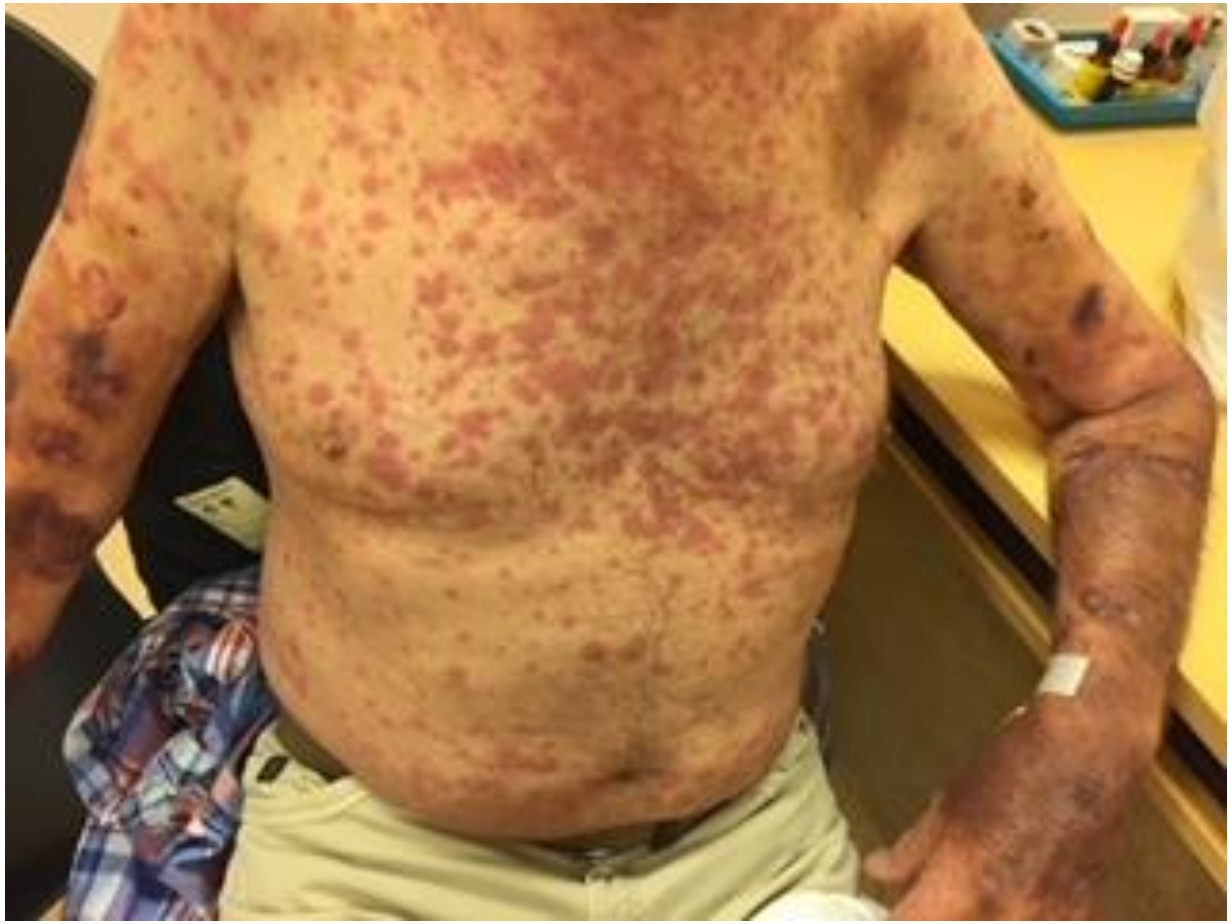
Humira

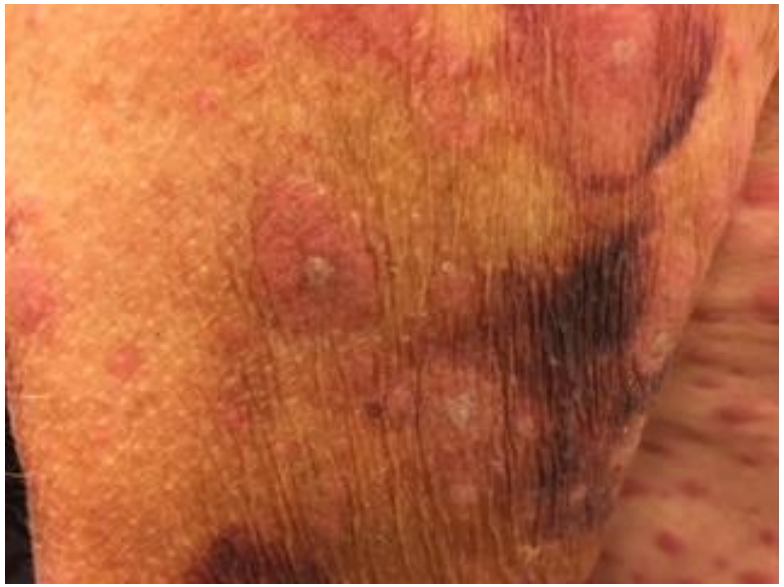
Sandimmun+Imurel

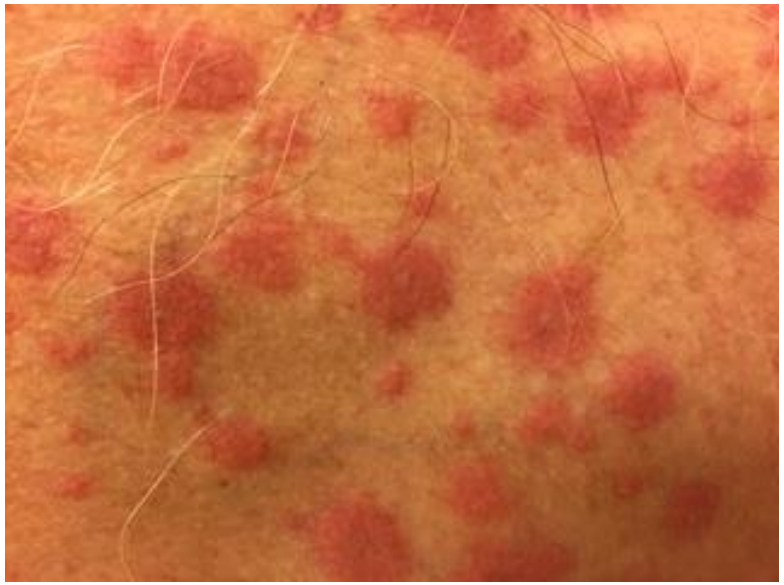
Sandimmun+CellCept

Sandimmun — IVIG 2018

Sweets syndrom
(febril neutrofil dermatose) 2017







Oslo universitetssykehus

Bilder med tillatelse fra ektefelle
Foto Ø.Midtvedt

2012	2013	2014	2015	2016	2017	2018	2019	2020

Klinikk:

Iridocyklitt
Redusert allmenntilstand
Feber
Brystsmerter

Kondritt øre
Diagnose: **Relapserende Polykondritt**

Sweets

Mors

Biokjemi:

SR 131,CRP 183,
Hb 9,9, MCV 96, sep-12 MCV>100

7,0

Blodtransfusjon hver 14/d
(EPO, G-CSF)

hver 7-10/d

Trombocytter 496

60

Revolade

Behandling:

Prednisolon 60mg/d – minste dose 17,5mg/d

Methotrexate

RoActemra

Kineret - utslett

Infliksimab

Humira

Sandimmun + Imurel

Sandimmun + CellCept

Sandimmun

IVIG 2018

2012	2013	2014	2015	2016	2017	2018	2019	2020

Klinikk:

Iridocyklitt — Kondritt øre — Sweets — Mors

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SR 131,CRP 183,

Hb 9,9, MCV 96 ,sep-12 MCV>100 — 7,0 — Blodtransfusjon hver 14/d (EPO, G-CSF) — hver 7-10/d

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Prednisolon 60mg/d – minste dose 17,5mg/d →

Methotrexate

RoActemra

Kineret - utslett

Infliksimab

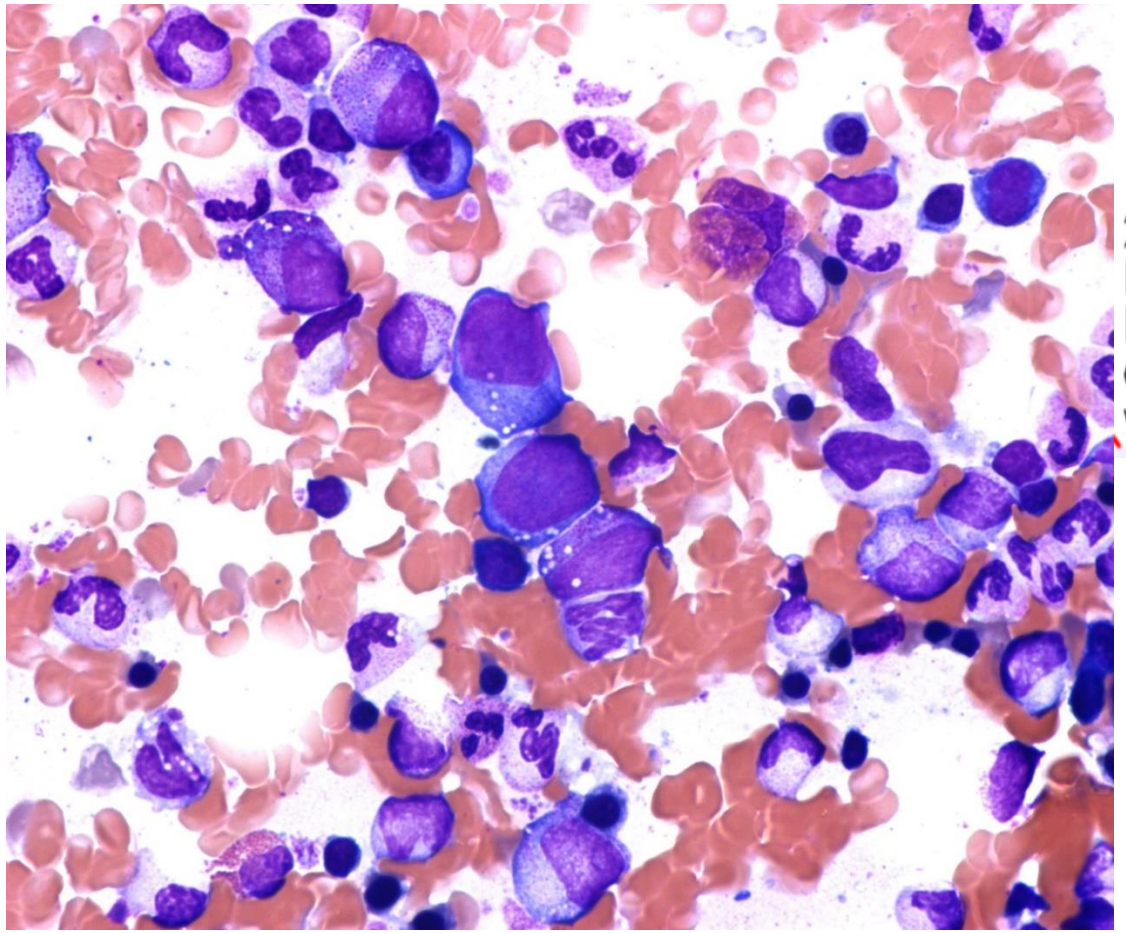
Humira

Sandimmun + Imurel

Sandimmun + CellCept

Sandimmun — IVIG 2018

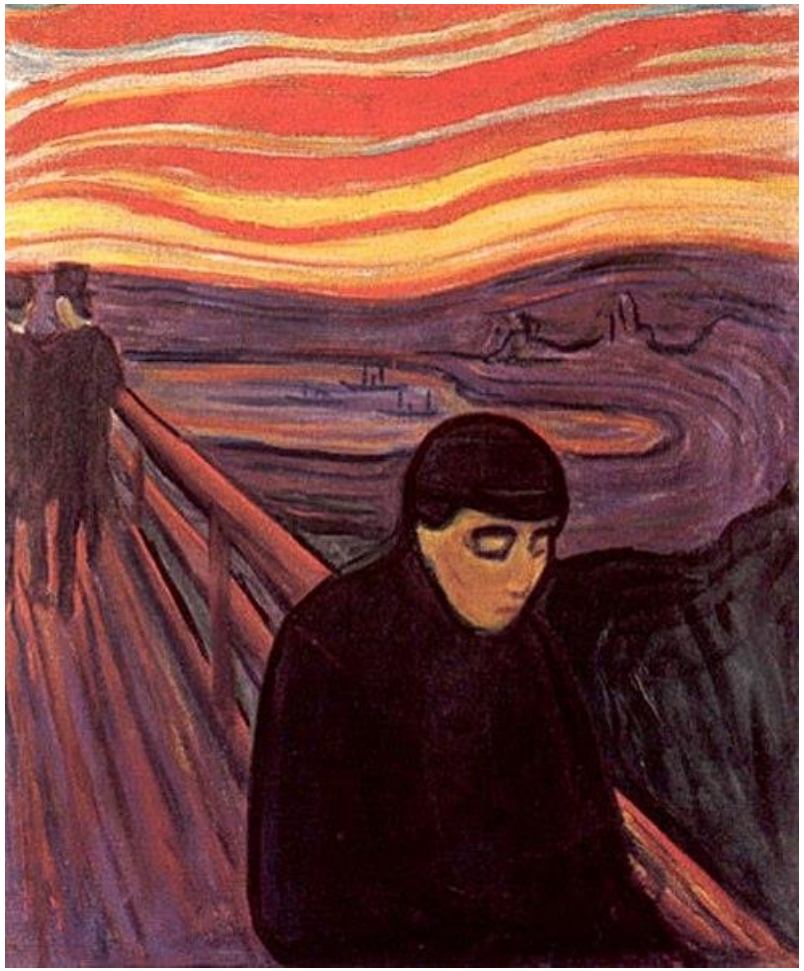
Vakuoler i myeloide og erytroide forløpere



2. BENMARGSUTSTRYK:
KVALITET: Optimal
BESKRIVELSE: Hyperplastisk, venstre forskjøvet erythropoiese med megaloblastisk trekk og dysplastiske forandringer (vakuolisering av cytoplasma av erythroblastene). Myelopoiesen viser også vakuolisering av promyelocytter, men viser ellers normal modning. Vi har ikke sett økt antall blaster.

Benmarg 12.04.15

Foto: Signe Spetalen, OUS



E. Munch, Fortvilelse (1894)
Foto Ø. Midtvedt

«Per» oppsummering:

Vedvarende inflammatorisk sykdom

Feber, redusert allmenntilstand, kondritt, iridocyklitt, Sweets syndrom

Nonkraniell KCA – relapserende polykondritt med hematologisk sykdom

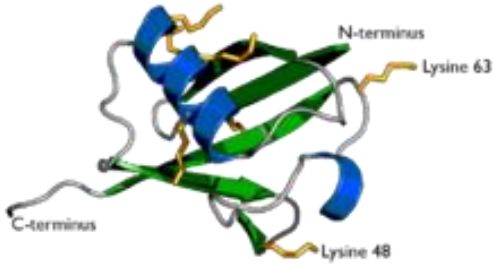
Høy SR, CRP, anemi, MCV > 100, trombocytopeni

Terapiresistent: Prednisolon > 20mg/d
(MTX, RoActemra, Kineret, Infliksimab, Humira, AZA, MMF, Cya, IVIG)

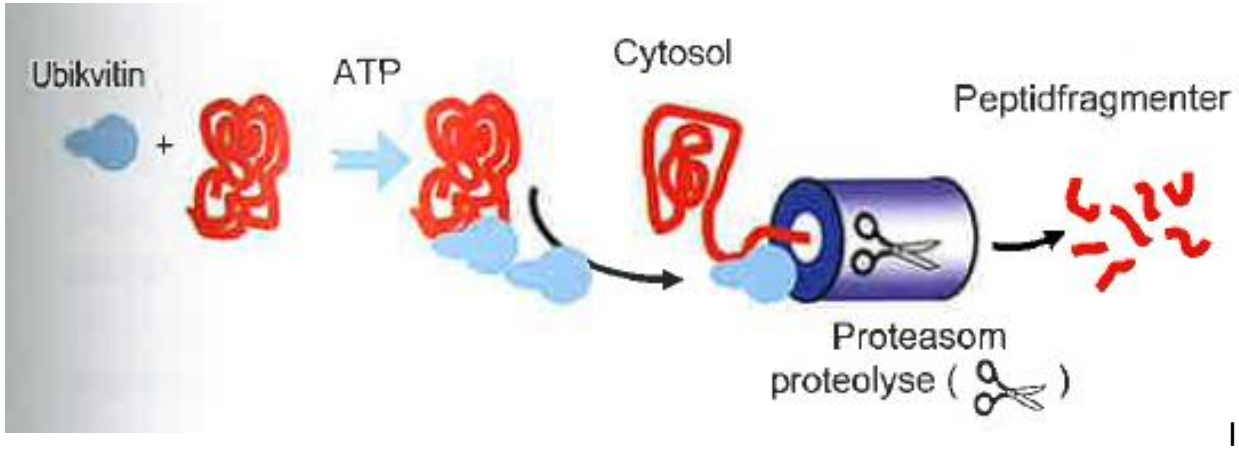
Benmargsbiopsi: -12,-13,-14,-15,-16,-19

Antall revmatologer: 34

Ubikvitin (ubiquitous = allestedsnærværende)



- Høyt konservert protein i alle eukaryote celler
- Oppdaget av Gideon Goldstein i 1975
- Ubikvitin = «Kiss of death»
- Ubikvitinering: Nobels pris i kjemi – 04, Aaron Ciechanover, Avram Hershko, Irwin Rose



Immunologi, B. Bogen, L.A. Munthe

Ubikvitinering

Infeksjonsmedisin

«Sars-Cov2/COVID-19 and its relationship with NOD2 and ubiquitinatin» Rivera et al Clin Immunol 2022

Endokrinologi

« Ubiquitin- proteasome system in diabetic retinopathy» Z.Svikle et al Peer 2022
«The role of ubiqitination and sumoylation in diabetic nephrotathy»
Gao C et al Biomed Res Int 2014

Nefrologi

«Ubiquitin, proteasomes and proteolytic Mechanisms activated by kidney disease»
Rajan V et al Bichim Biophys acta 2008

Hematologi

« Ubiquitination and Ubiquitin –like modifications in Multiple Myeloma: Biology and Therapy « Wirth M. Cancers 2020

Gastroenterologi

«Ubiquitin-specific proteases in inflammatory bowel disease-related signalling pathway regulation»
R.Chen et al Cell Death Dis 2022
«Ubiquitin-proteasome system and oxidative stress in liver transplantation»,
Alva N et al World J gastroenterol 2018

Revmatologi

«Post-translation modifications in T cells in systemic erythematosus lupus»
Fang F et al Rheumatology 2021
«The ubiquitin proteasome system as a potenial therapeutic target for systemic sclerosis» Meiners S et al Transl res 2018

Dermatologi

«Ubiquitination in melanoma pathogenesis and treatment», J- M et al Cancer med 2017
«Ubiquitination – proteasome system: A new player in the pathogenesis of psoriasis and clinical implications», L. Yang et alk J Dermatol Sci 2018

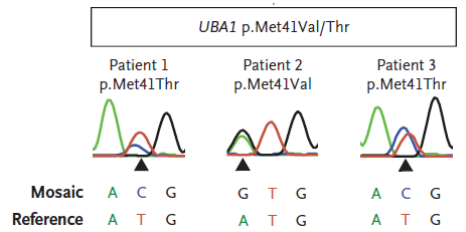
The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

Somatic Mutations in *UBA1* and Severe Adult-Onset Autoinflammatory Disease

D.B. Beck, M.A. Ferrada, K.A. Sikora, A.K. Ombrello, J.C. Collins, W. Pei, N. Balanda, D.L. Ross, D. Ospina Cardona, Z. Wu, B. Patel, K. Manthiram, E.M. Groarke, F. Gutierrez-Rodrigues, P. Hoffmann, S. Rosenzweig, S. Nakabo, L.W. Dillon, C.S. Hourigan, W.L. Tsai, S. Gupta, C. Carmona-Rivera, A.J. Asmar, L. Xu, H. Oda, W. Goodspeed, K.S. Barron, M. Nehrebecky, A. Jones, R.S. Laird, N. Deuitch, D. Rowczenio, E. Rominger, K.V. Wells, C.-C.R. Lee, W. Wang, M. Trick, J. Mullikin, G. Wigerblad, S. Brooks, S. Dell'Orso, Z. Deng, J.J. Chae, A. Dulau-Florea, M.C.V. Malicdan, D. Novacic, R.A. Colbert, M.J. Kaplan, M. Gadina, S. Savic, H.J. Lachmann, M. Abu-Asab, B.D. Solomon, K. Retterer, W.A. Gahl, S.M. Burgess, I. Aksentijevich, N.S. Young, K.R. Calvo, A. Werner, D.L. Kastner, and P.C. Grayson

NEJM, oct -20



- Gensekvensering av pasienter med uklar diagnose
- 25 menn med mutasjon i UBA1
- Ingen familiemedlemmer med samme mutasjon
- Ingen friske med samme mutasjon
- Knockout UBA1 gen hos sebrafisk ga inflammasjon

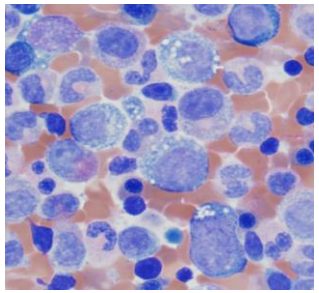


Foto Pinterest.com

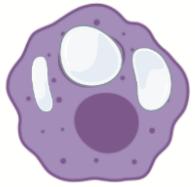
Table 1. Demographic and Clinical Characteristics of Participants with the VEXAS Syndrome.*

Characteristic	Participants (N=25)
Demographic characteristics	
Male sex — no. (%)	25 (100)
Median age at onset (range) — yr	64 (45–80)
Died before the current study — no. (%)	10 (40)
Genetic characteristics	
Somatic *UBA1* (NM_003334.3) variant (p.Met41) — no. (%)	25 (100)
p.Met41Thr (c.122T→C)	15 (60)
p.Met41Val (c.121A→G)	5 (20)
p.Met41Leu (c.121A→C)	5 (20)
Key clinical features	
Fever — no. (%)	23 (92)
Skin involvement — no. (%)†	22 (88)
Pulmonary infiltrate — no. (%)	18 (72)
Ear and nose chondritis — no. (%)	16 (64)
Venous thromboembolism — no. (%)	11 (44)
Macrocytic anemia — no. (%)	24 (96)
Bone marrow vacuoles — no./total no. (%)	18/18 (100)
Laboratory findings	
Median C-reactive protein (IQR) — mg/liter	73 (18–128)
Median ESR (IQR) — mm/hr	97 (64–124)
Current or past treatment	
Glucocorticoids — no. (%)	25 (100)
Median no. of synthetic DMARDs (IQR)	2 (1–3)
Median no. of biologic or target synthetic DMARDs (IQR)	2 (0.5–3)
Diagnostic or classification criteria that were met — no. (%)	
Relapsing polychondritis	15 (60)
Sweet's syndrome	8 (32)
Myelodysplastic syndrome	6 (24)
Multiple myeloma or monoclonal gammopathy of undetermined significance	5 (20)
Polyarteritis nodosa	3 (12)
Giant-cell arteritis	1 (4)

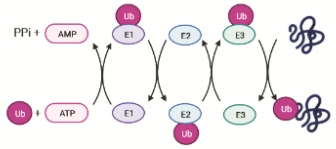
* DMARDs denotes disease-modifying antirheumatic drugs, ESR erythrocyte sedimentation rate, IQR interquartile range, and p.Met41 methionine-41.
† The most common skin-biopsy findings were neutrophilic dermatosis (in 8 participants), leukocytoclastic vasculitis (in 7 participants), and medium-vessel arteritis (in 3 participants).

Identifying VEXAS

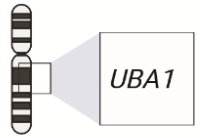
V **Vacuoles:**
Vacuolated myeloid and erythroid precursors



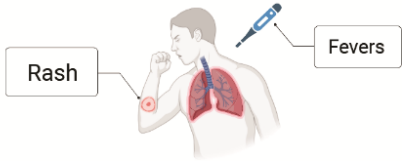
E **E1-Enzyme:**
Mutations in *UBA1* lead to lack of cytoplasmic E1 enzyme



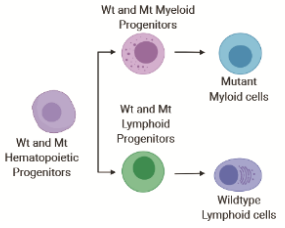
X **X-linked:**
UBA1 gene located on X chromosome



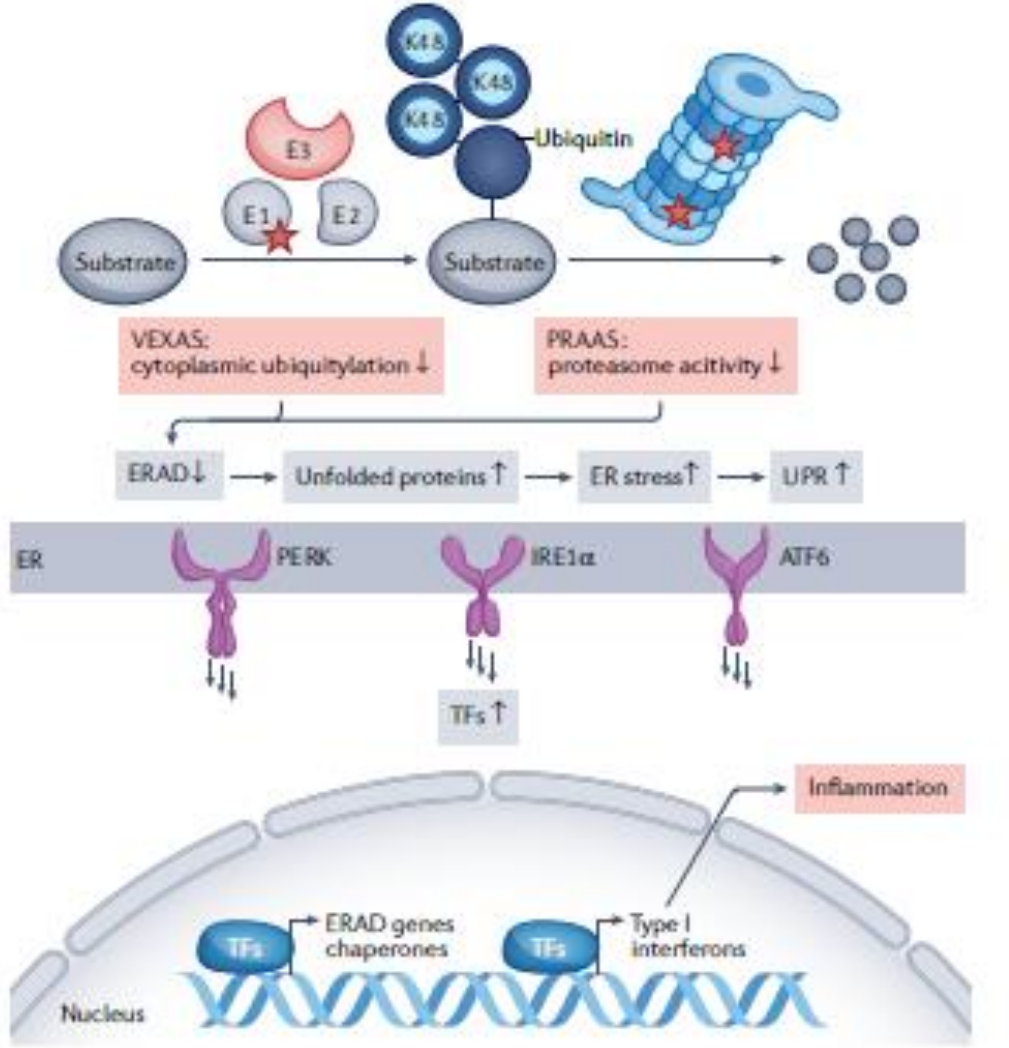
A **Autoinflammatory:**
Severe inflammation, steroid dependent



S **Somatic:**
Mutations in *UBA1* restricted to myeloid origin cells

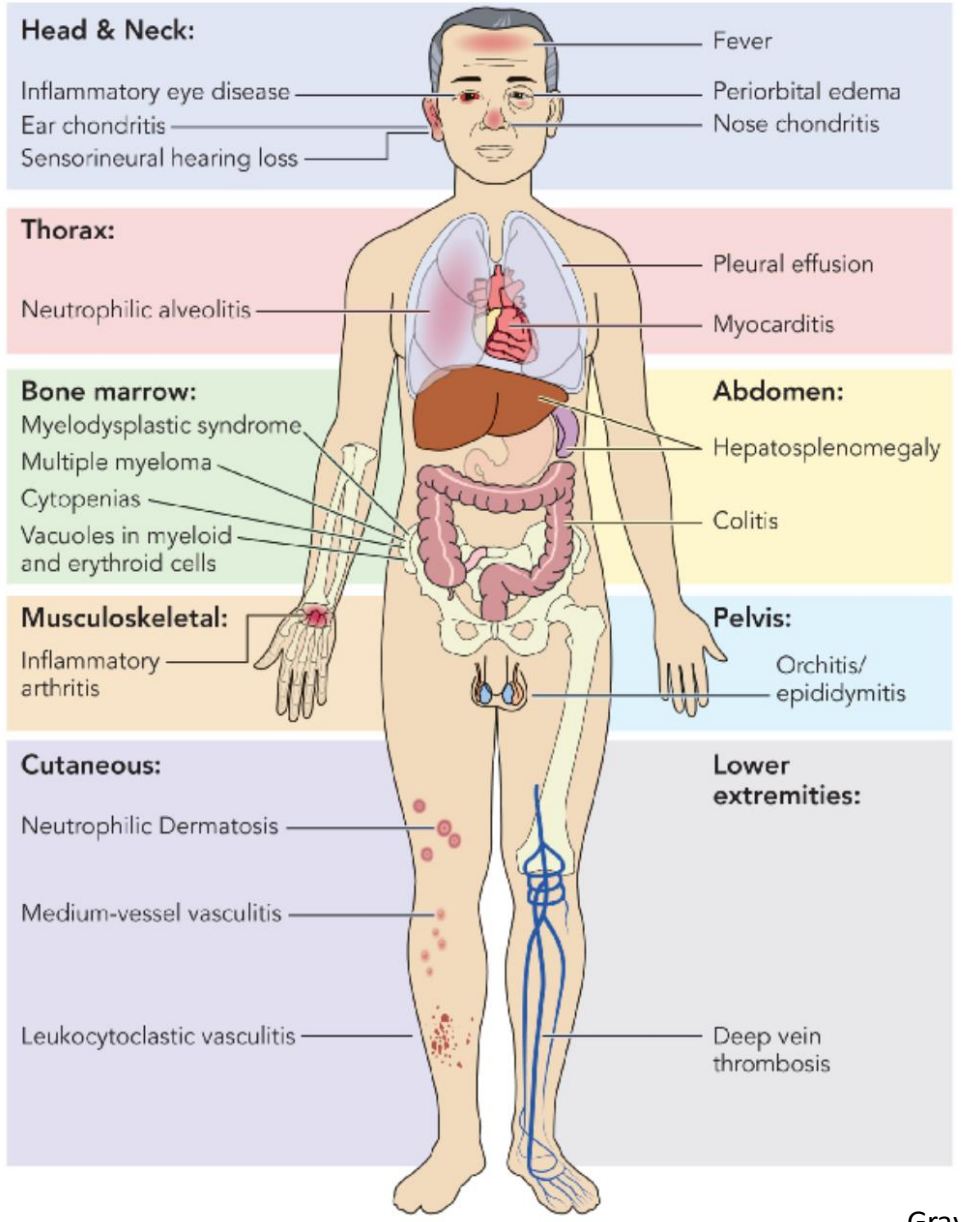


Patofysiologi



Disorder of ubiquitylation: unchained inflammation
D Beck et al Nature Reviews Rev Aug22

VEXAS symptomer



Nyre

«Acute tubulointerstitial nephritis revealing VEXAS syndrome»
Broek et al Kidney International 2022

Grayson PC et al. Blood, 2021

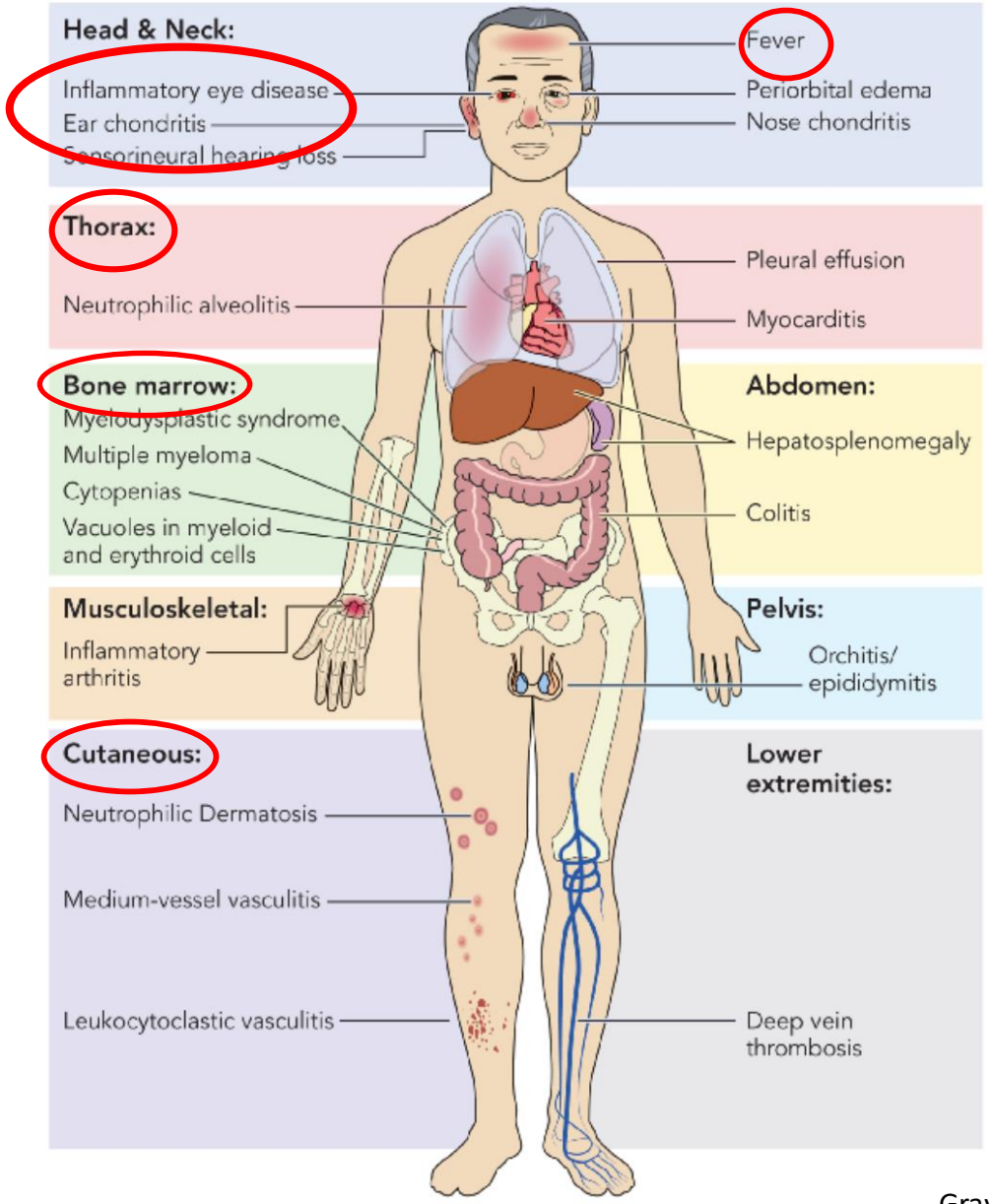
«Per» og VEXAS symptomer

»Per» var registrert i NOSVAR
(Norsk systemisk bindevevssykdom og vaskulitt register)
Prøve fra 2012 sendt til genetisk undersøkelse

2019 Prof.G.E.Tjønnfjord:
Gentest mtp Shwachman - Diamond syndrom: Negativ
Reanalysert

Diagnose:
Genmutasjon UBA1 gen: Met41Thr

www.genetikkportalen.no



Grayson PC et al. Blood, 2021

Nyre

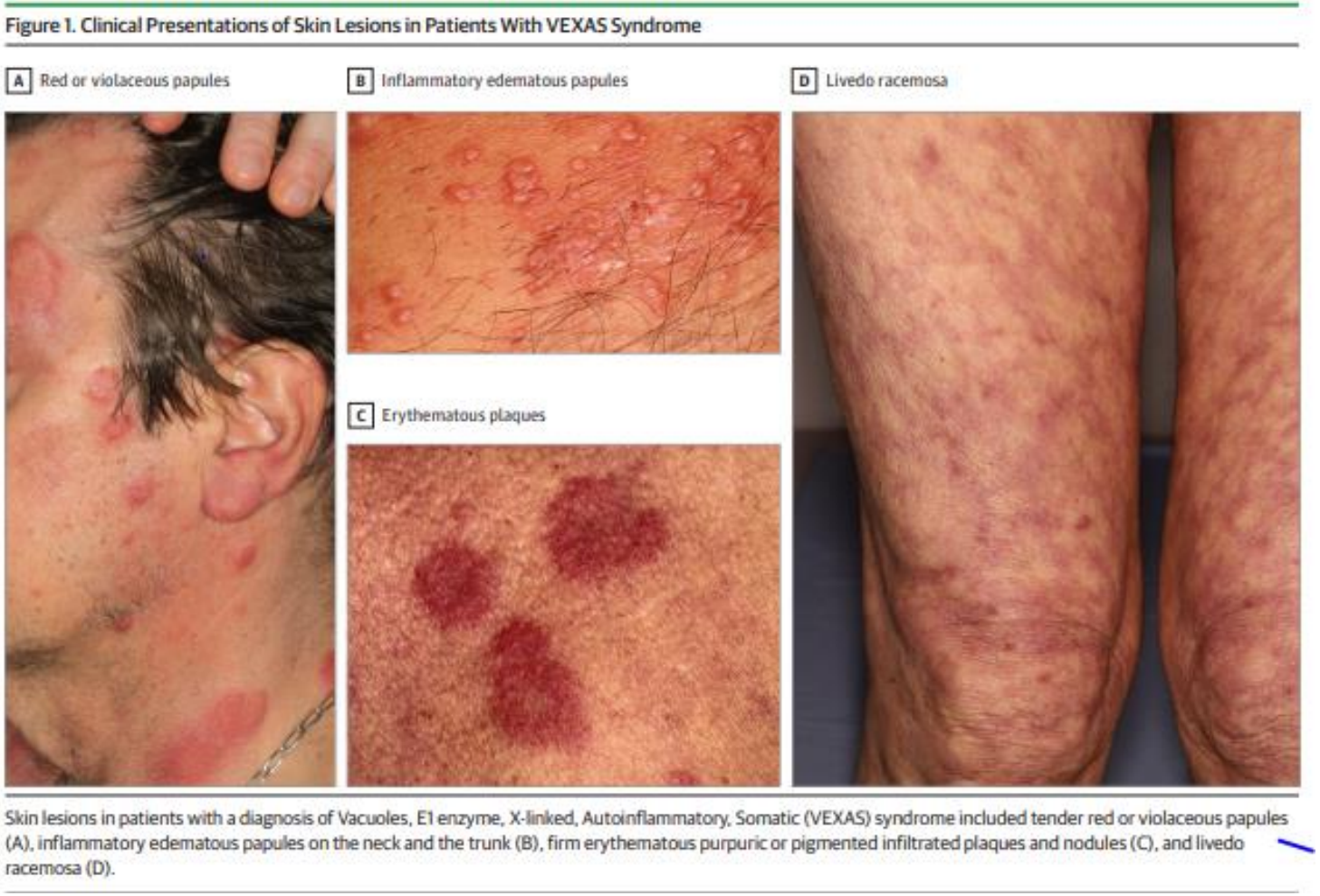
«Acute tubulointerstitial nephritis revealing VEXAS syndrome»
Broek et al Kidney International 2022

VEXAS og hud

Vanlig (88%)
Sweets liknende utslett
Noduli
Livedo racemosa

Kineret, ofte hudreaksjon

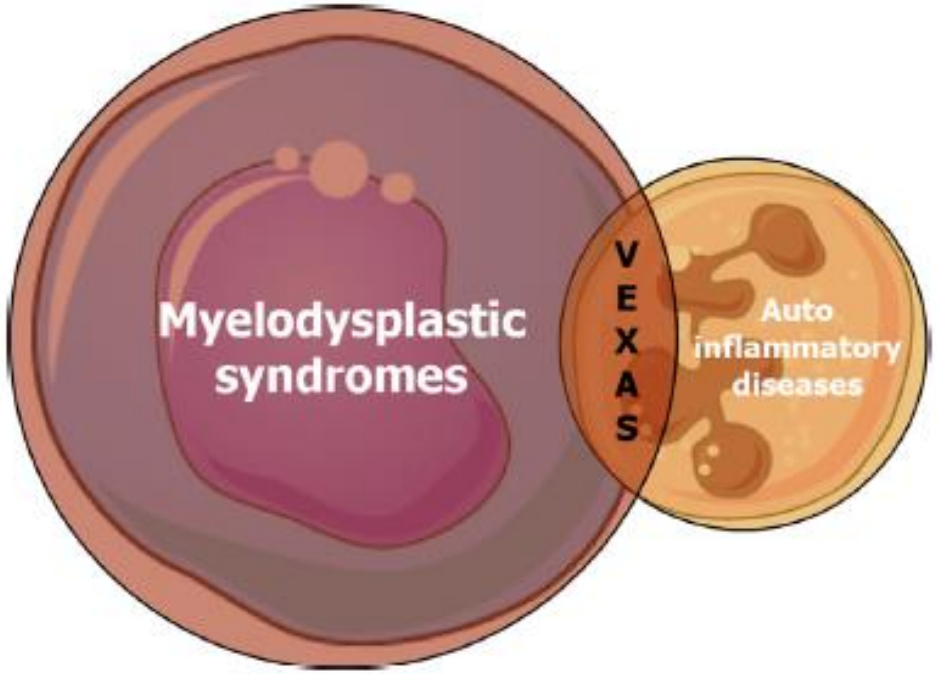
Histologi:
Nøytrofil dermatose
Leukocytoklastisk vaskulitt
CD68+ celler



UBA1 variations in Neutrophilic Dermatosis Skin Lesions of Patients with VEXAS Syndrome,
E.Zakine et al ,JAMA Dermatology 2021
VEXAS syndrome for the dermatologist
D. Sterling et al J Am Acad Dermatol 2022
Somatic mutations in UBA1 and severe Adult-onset Autoinflammatory disease,
D.B.Beck et al NEJM-20

VEXAS og hematologi

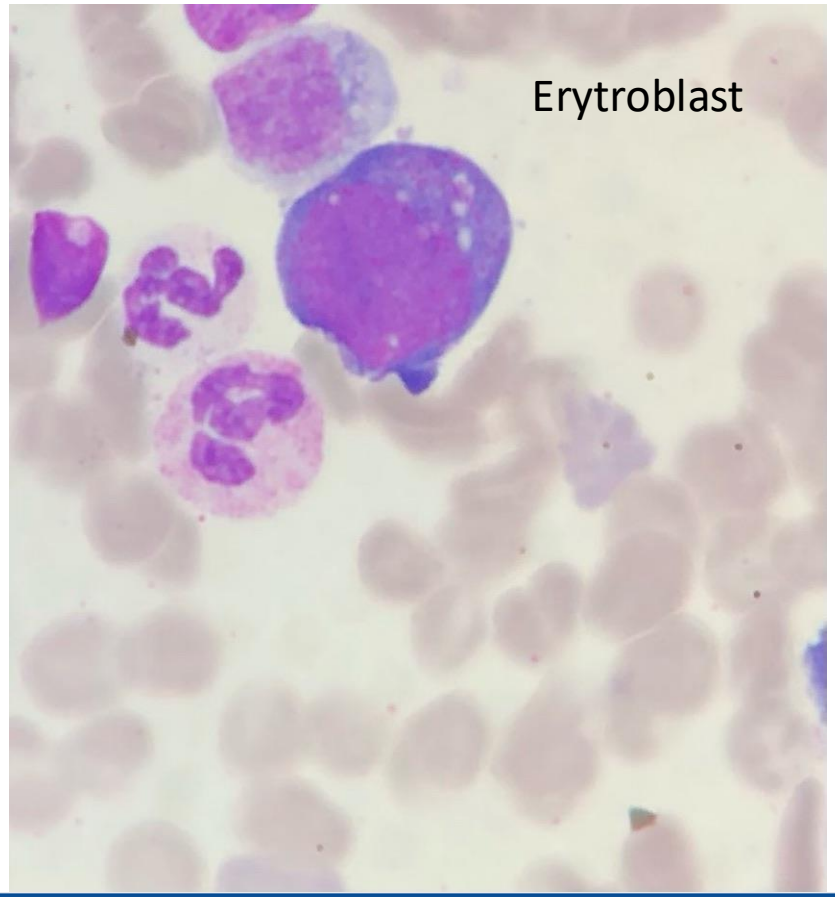
- Makrocytær anemi (96%)
- Trombocytopeni
- Benmarg; vakuoler (18 av 18 pas)
- Myelodysplastisk syndrom (24%, 50% [x])
- Monoklonal gammopati (20%)
- Trombose (44%)



A Oganesyam , Seminars in hematology 2021

X) Further characterization of clinical and laboratory features in VEXAS syndrome:
Large-scale analysis of a multicenter case series of 116 French patients. Georgin-Lavialle Br J Derm2022
Somatic mutations in UBA1 and severe Adult-onset Autoinflammatory disease, D.B.Beck et al NEJM-20

Vakuoler er ikke spesifikt for VEXAS



Mann f-58:
MDS fra 2015
Kineret 200 mg/d 2018
Ikke påvist mutasjon i UBA1
Allogen tx jan-23

Årsaker til vakuoler:
MDS
Alkohol overforbruk
Sink forgiftning
Kobber mangel

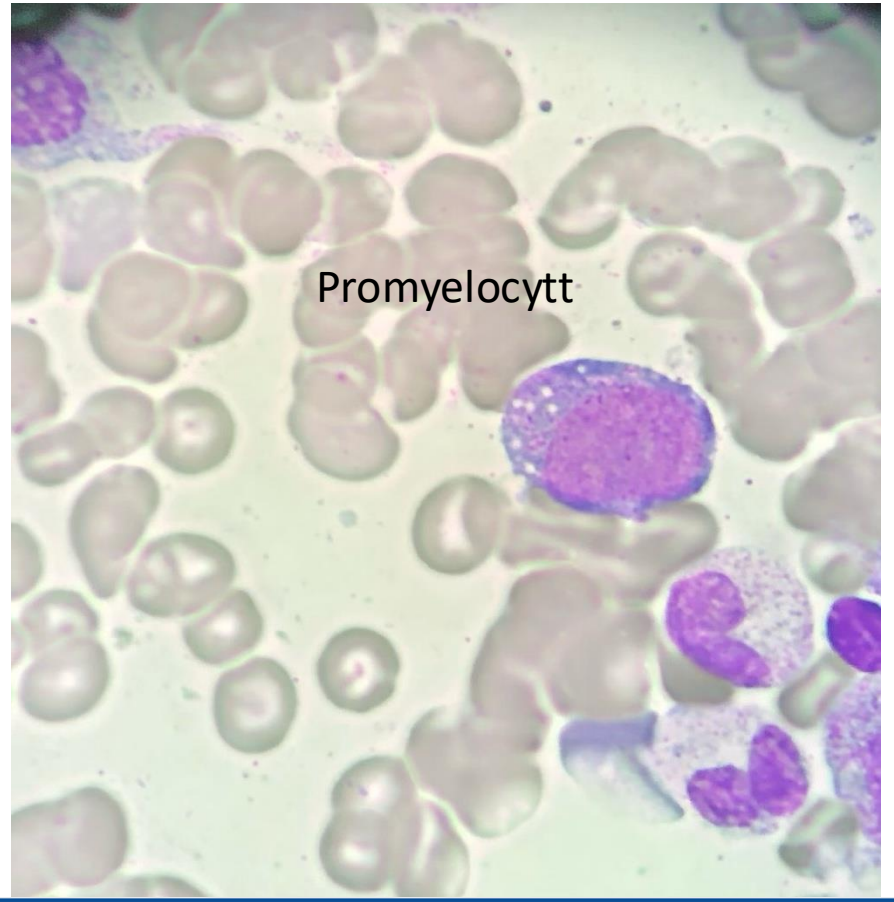


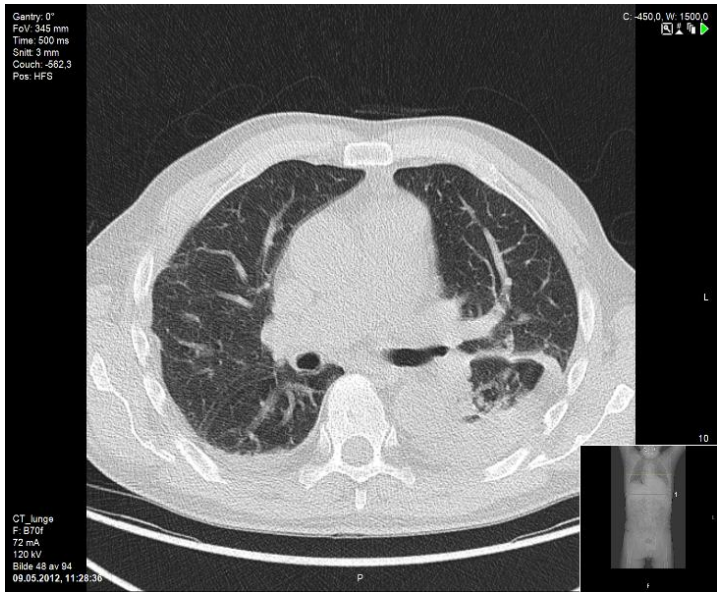
Foto: Hematolog Moksnes ved SIV. Med tillatelse fra pasient og Moksnes.

VEXAS og infeksjonsmedisin

- Feber
- Autoinflammatorisk syndrom
- Opportunistisk infeksjon
- Obs oppbluss vs infeksjon

VEXAS og lunge

Lungeinfiltrat er vanlig (72%)



Somatic mutations in UBA1 and severe Adult-onset Autoinflammatory disease,
D.B.Beck et al NEJM-20

Pleuropulmonary manifestation of Vacuoles, E 1 enzyme, X-Linked, Autoinflammatory, Somatic (VEXAS) syndrome
R Borie et al Chest -22

TABLE 3] Pulmonary CT Scan Characteristics of Patients With VEXAS Syndrome With Pulmonary Involvement

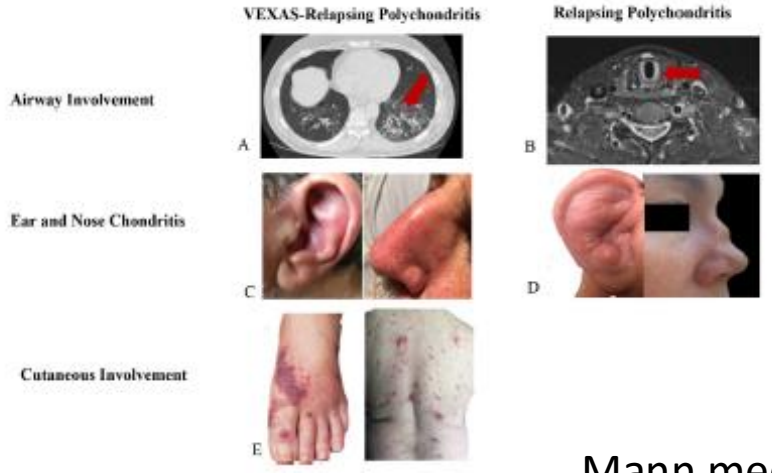
CT Scan Abnormalities	Initial (n = 45)	Follow-up (n = 81)
Parenchymal disease	45 (100)	73 (90)
Ground-glass opacities	39 (87)	60 (74)
Consolidations	22 (49)	42 (52)
Reticulations	17 (38)	23 (28)
Septal lines	23 (51)	46 (57)
Fibrosis	1 (2)	1 (1)
Nodules	21 (47)	20 (25)
Micronodules	17 (38)	20 (25)
Pleural effusion	24 (53)	37 (46)
Small	23 (96)	28 (34)
Unilateral	14 (58)	21 (26)
Right	9	15
Left	5	6
Pericardial effusion	7 (15)	11 (13)
Mediastinal adenomegaly	26 (58)	53 (65)
< 20 mm	24 (92)	47 (88)
Multiple	19 (73)	35 (66)
Cluster		
1[a]	15 (33)	21 (26)
2[b]	18 (40)	37 (46)
3[c]	13 (28)	18 (22)
Extent of abnormalities		
< 10%	21 (47)	36 (44)
10%-20%	10 (22)	16 (19)
20%-30%	8 (18)	14 (17)
30%-40%	3 (7)	5 (6)
40%-50%	3 (7)	2 (2)
> 50%	0 (0)	2 (2)
Normal	0	5 (5)

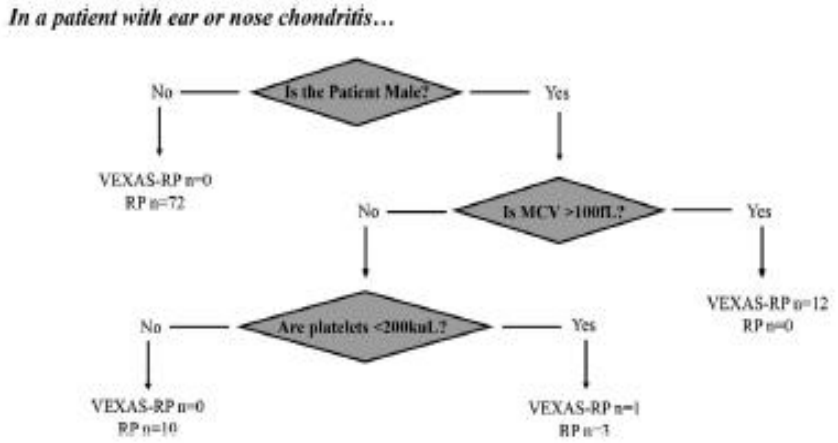
Data are presented as No. (%) or No. VEXAS = vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic.
[a]Suggests organising pneumonia or infection.
[b]A nonspecific pattern.
[c]Suggests heart failure.

VEXAS og revmatologi

- Relapserende polykondritt (88%)
- PAN (12%)
- KCA (4%)
- Andre;

  SLE, ANCA vaskulitt, SpA





Mann med kondritt + MCV>100 + trombocytter<200 = VEXAS
100% sensitivitet og 96 % spesifisitet

Somatic mutations in UBA1 define a distinct subset of relapsing Polychondritis patients with VEXAS, MA Ferrada et al AR okt-21

Somatic mutations in UBA1 and severe Adult-onset Autoinflammatory disease, D.B.Beck et al NEJM-20

Forekomst

- Alder > 50 år:
- Menn 1:4269, (232 i Norge)
- Kvinner 1:26238, (39 i Norge)
- VEXAS syndrome in a woman, T.Barba et al Rheumatol -21

Estimated prevalence and clinical manifestation of UBA1 Variants associated with VEXAS syndrome in a clinical population
Beck et al , JAMA 2023

Vexas ved OUS

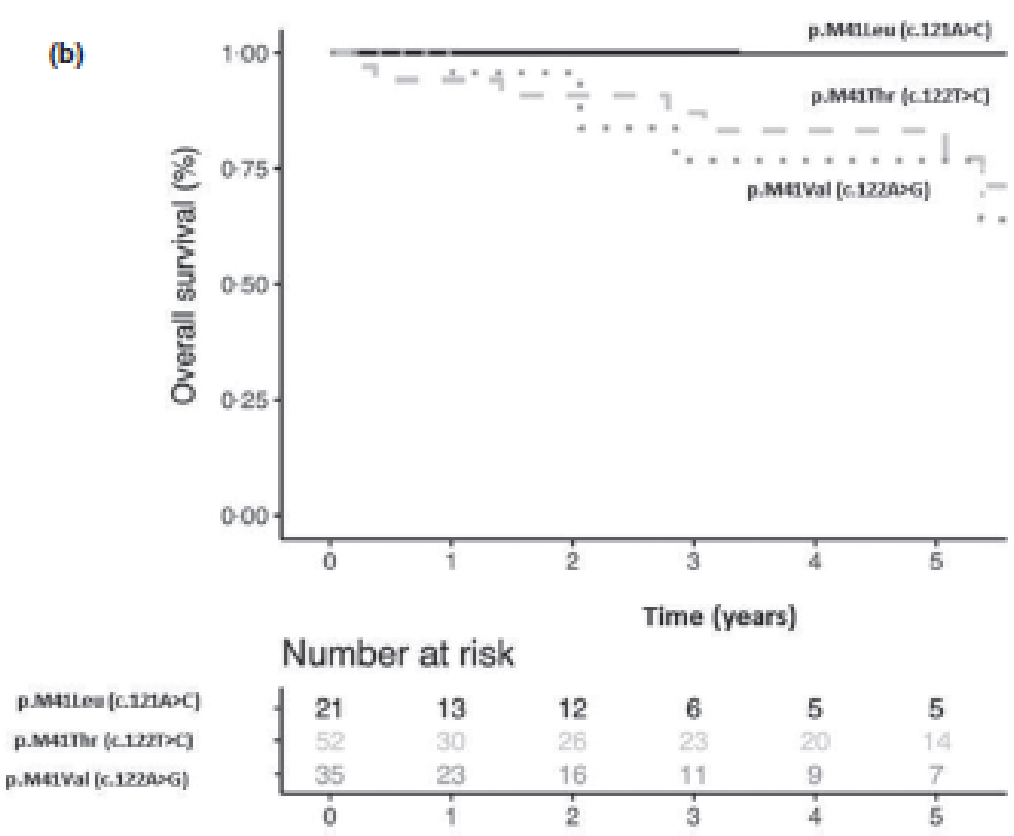
Menn	16*
Alder ved debut	44 - 81 år (median 70)
UBA1 mutasjon	Met41Thr(7) Met41Val (5) Met41Leu (3) C118-1G>C**
Feber Utslett Kondritt Lunge MCV>100 Trombocytopeni	12 12 8 9 15 6
Behandling	Prednisolon 16, Jakavi 3, Plaquenil 1, RoActemra 4, Kineret 4, Methotrexate 3, Infliksimab 2, Imurel 2, CellCept 2, IVIG 2, Rituximab 1, Sendoxan 1, Humira 1, Sandimmun 1

* 2 diagnose post mortem
  44 år gammel mann fra Island innlagt OUS i 2018
  3 mors etter diagnose tidspunkt

** «Atypical splice-site mutations causing VEXAS Syndrome»,
O.Hermine, Rheumatology 2021

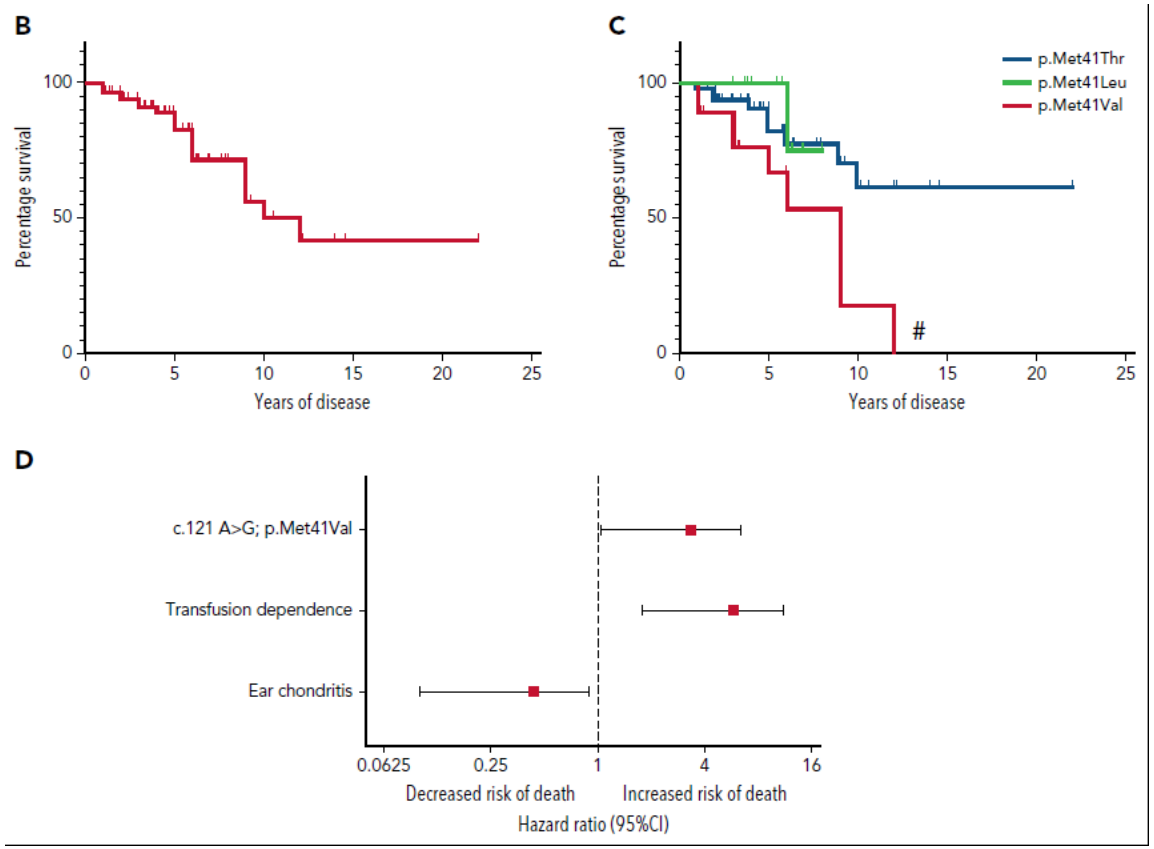
Mortalitet

Frankrike 116 VEXAS



5 års overlevelse på 63%

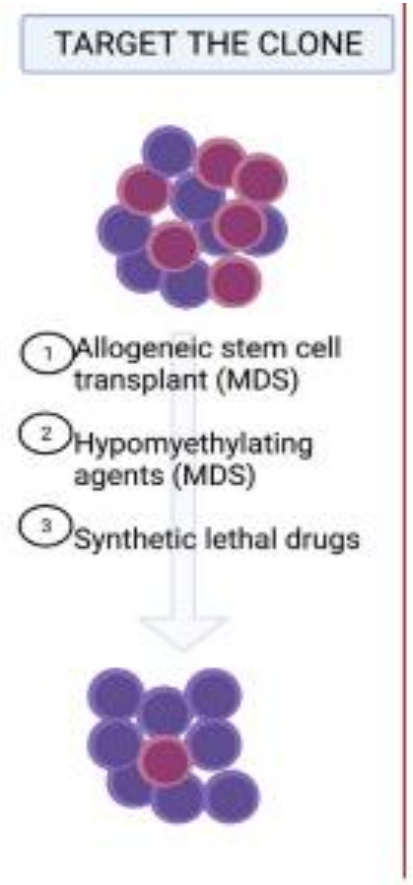
US-UK 83 VEXAS



Translatation of cytoplasmic UBA1 contributes to VEXAS syndrome pathogenesis, M.A. Ferrada et al Blood sep-22
Further Characterixation of clinical and laboratory features in VEXAS syndrome: large-scale analyses of a multicentre Case series of 116 French patiens, S. Georgin-Lavialle et al BJD 2021

Oslo universitetssykehus

Behandling



Retrospektiv 6 pasienter med VEXAS syndrom (46-65 år ved behandling)
3 remisjon (32, 37 og 38 mnd etter behandling)
2 remisjon (3 og 5 mndr etter behandling)
1 behandlingsrelatert død

Successful allogeneic haematopoietic stem cell transplantation in patients with VEXAS syndrome: A 2- center experience
A.Diarra, Blood Advances oct-21

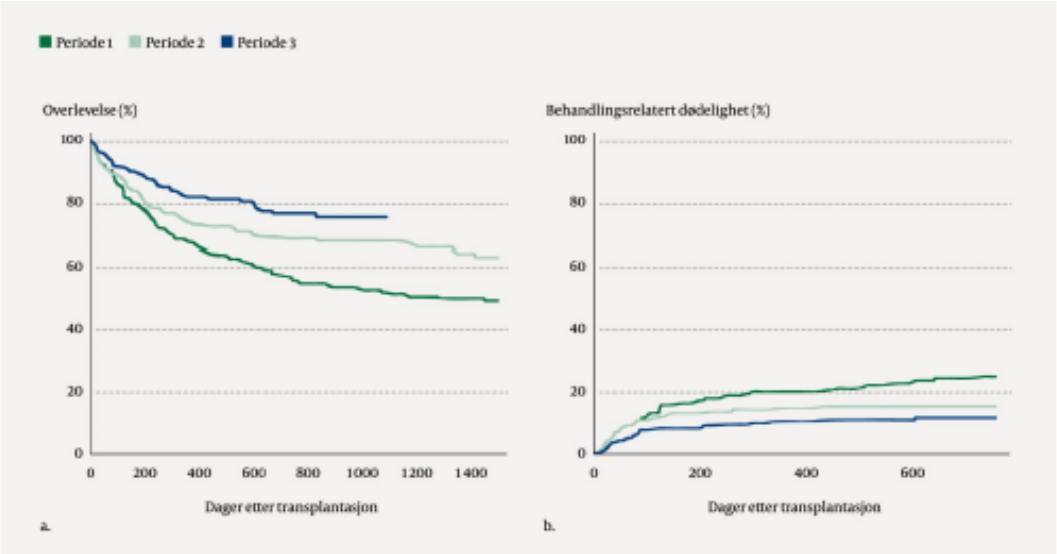
A Phase II study of allogenic haematopoietic stem cell transplant for subjects with VEXAS syndrome. NIH, clinicaltrials.gov

Azacitidin ; 9 av 12 med komplett eller partiell effekt

Phase II prospective trial of azacitidine in steroid-dependent or Refractory systemic autoimmune/inflammatory disorders and VEXAS Syndrome associated with MDS and CMML, Mekinian A et al Leukemia 2022

Toward a pathophysiology inspired treatment of VEXAS syndrome, M. Heiblig et al , Seminars in Hematology 2021

Allogen stamcelletx ved OUS



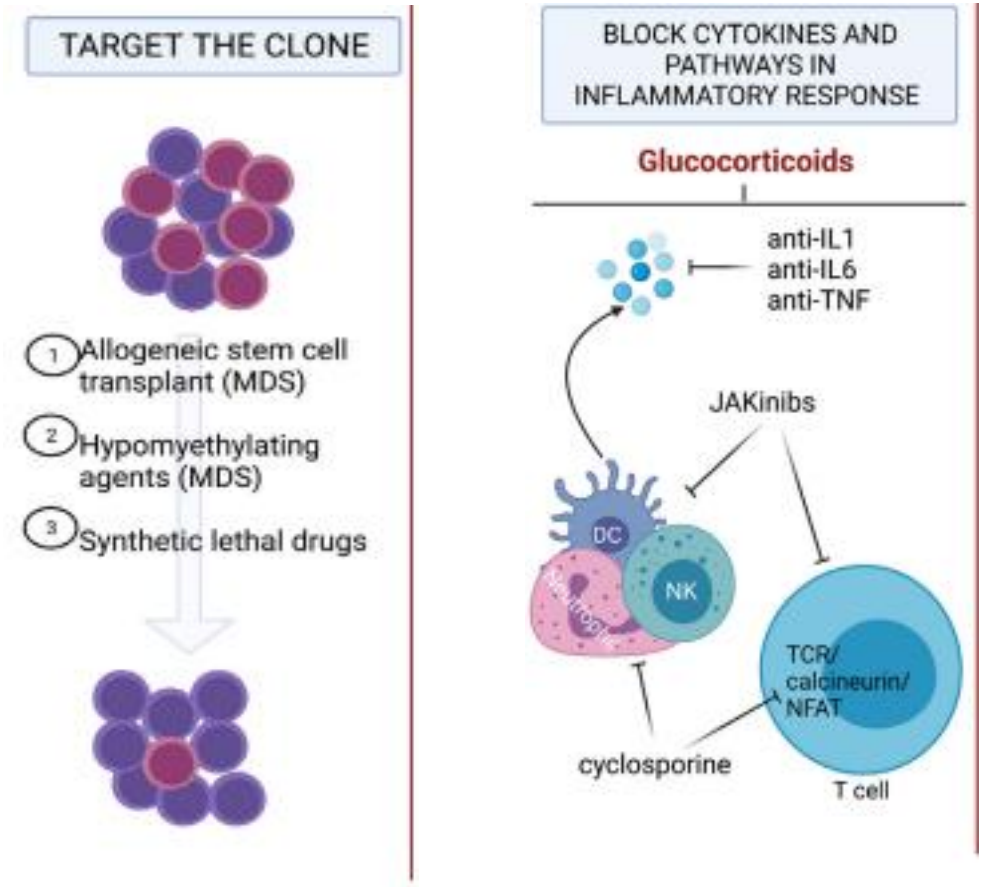
Figur 2A, 2B Kliniske resultater etter allogen stamcelletransplantasjon i 2015–21. Resultater for a) overlevelse og b) behandlingsrelatert dødelighet. Mørkegrønn linje angir periode 1 (n = 186), lysegrønn linje periode 2 (n = 177) og blå linje periode 3 (n = 226). Periode 3 i figur 2a har kortere observasjonstid enn periode 1 og 2.

Pasient- og donorkarakteristika for voksne pasienter behandlet med allogen stamcelletransplantasjon ved Avdeling for blodsykdommer ved Oslo universitetssykehus fra og med 24.5.2015 til og med 23.5.2021.

Variabel	Hele perioden 24.5.2015–23.5.2021 (2 192 dager)	Periode 1 24.5.2015–23.5.2017 (731 dager)	Periode 2 24.5.2017–23.5.2019 (730 dager)	Periode 3 24.5.2019–23.5.2021 (731 dager)
Antall pasienter	589	186	177	226
Alder ved transplantasjon, år, median (spredning)	57 (15–74)	56 (16–71)	55 (15–74)	58 (16–74)
Kjønn				
Kvinner	225	67	61	97
Menn	364	119	116	129
Grunnsykdom, antall (%)				
Akutt leukemi	291 (49,4)	93 (50,0)	89 (50,3)	119 (52,7)
Myelodysplastisk syndrom / myeloproliferative sykdommer	161 (27,3)	47 (25,3)	53 (29,9)	61 (27,0)
Lymfom / kronisk lymfatisk leukemi	91 (15,4)	39 (21,0)	24 (13,6)	28 (12,4)
Kronisk myelogen leukemi	11 (1,9)	3 (1,6)	2 (1,1)	6 (2,7)
Ikke-malign sykdom	24 (4,1)	4 (2,2)	8 (4,5)	12 (5,3)

Allogen stamcelletransplantasjon hos voksne 2015-21
Camilla Dao Vo et al Tidsskriftet 2023

Behandling



Toward a pathophysiology inspired treatment of VEXAS syndrome, M. Heiblig et al , Seminars in Hematology 2021

TO THE EDITOR:

Ruxolitinib is more effective than other JAK inhibitors to treat VEXAS syndrome: a retrospective multicenter study

Maël Heiblig,[1] Marcela A. Ferrada,[2,*] Matthew T. Koster,[3,*] Thomas Barba,[4,*] Mathieu Gerfaud-Valentin,[5] Arsène Mékinian,[6] Henrique Coelho,[7] Gaelle Fossard,[1] Fiorenza Barraco,[1] Lionel Galicier,[8] Boris Bienvenu,[8] Pierre Hirsch,[9] Guillaume Vial,[10] Anne Blandine Boutin,[11] Joris Galland,[12] Guillaume Le Guenno,[13] Adrien Bigot,[14] Kenneth J. Warrington,[3] Tanaz A. Kermani,[15] Peter C. Grayson,[2] Bhavisha A. Patel,[16] David B. Beck,[17,18] Yvan Jamilloux,[5,†] Pierre Fenaux,[19,†] and Pierre Sujobert[20]

Blood aug-22

Klinisk respons

Biologisk respons

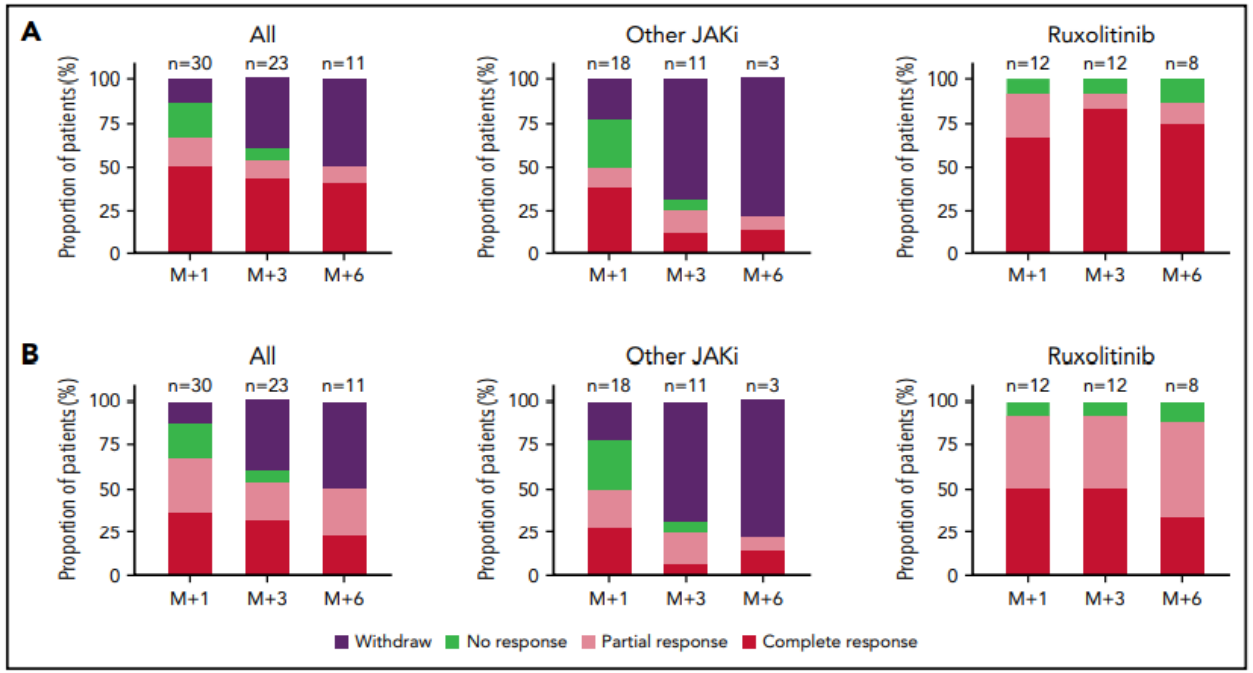
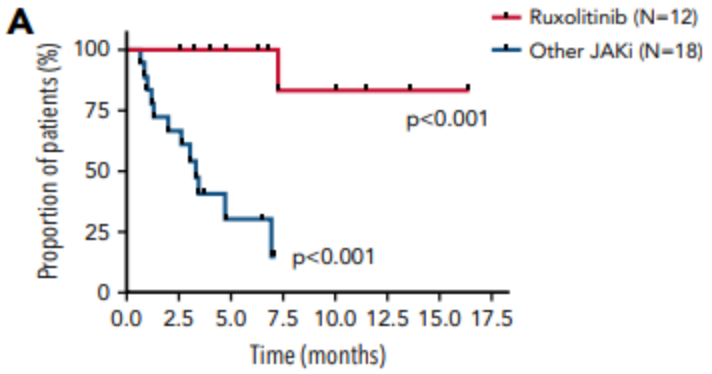
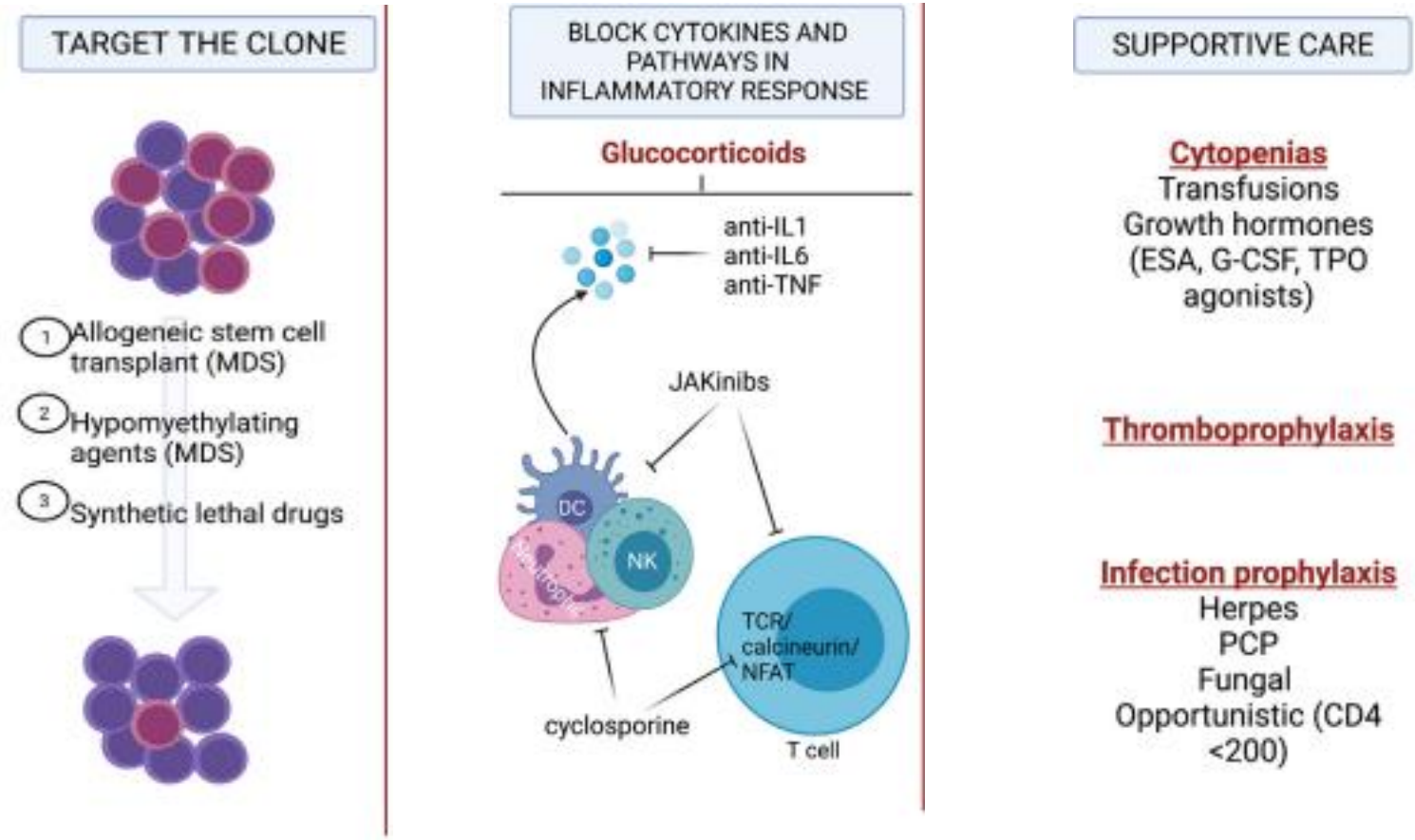


Figure 1. Antiinflammatory effects of JAKi in patients with VEXAS syndrome. (A) Overall clinical response rates of patients treated with JAKi during 1, 3, or 6 months; results are presented for the whole cohort (left), for patients receiving other JAKi than ruxolitinib (middle), or for patients receiving ruxolitinib (right). (B) Overall biological response rates of patients treated with JAKi during 1, 3, or 6 months; results are presented for the whole cohort (left), for patients receiving other JAKi than ruxolitinib (middle), or for patients receiving ruxolitinib (right).

30 pasienter med VEXAS
Ruxolitinib 12
Tofacitinib 11
Baricitinib 4
Upadacitinib 3



Behandling



Toward a pathophysiology inspired treatment of VEXAS syndrome, M. Heiblig et al , Seminars in Hematology 2021

Hva sa han egentlig ?...

- VEXAS – «hemato autoinflammatorisk» syndrom
- Stor imitator
- Vurder VEXAS ved uklar inflammasjon hos menn med makrocytær anemi
- Diagnose: Genetisk mutasjon i UBA1 gen (genetikkportalen.no)
- Behandling i samarbeid med hematolog; Prednisolon, Jakavi
- Allogen stamcelletransplantasjon hos selekterte pasienter?

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Foto pinterest.com