Interaktivna predstavitev posterjev in odmor za kavo, Thursday, May 15 2025, 15:30-17:00
Location: avla
Session: **Interaktivna predstavitev posterjev / Interactive poster session**
Chairs: Mojca Urbančič and Alenka Lavrič Groznik

PO-15
Bilateral panscleritis in a patient with relapsing polychondritis and VEXAS syndrome
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This case presentation aims to discuss a patient with bilateral panscleritis, relapsing polychondritis, and heart arrhythmias, in whom a rare somatic mutation in the UBA1 gene was identified. Based on the clinical presentation and diagnostic investigations, the patient met the criteria for VEXAS syndrome. The acronym VEXAS refers to V: vacuoles, often seen in bone marrow biopsy cells, E: E1-ubiquitin-activating enzyme, encoded by the UBA1 gene, X: the UBA1 gene is located on the X chromosome, A: autoimmune inflammation, and S: somatic, non-inherited mutations.
A 55-year-old male was referred to the ophthalmology clinic due to worsening symptoms of bilateral red, painful eyes, and blurred vision. Clinical examination, along with ultrasonography, revealed significant inflammation of the entire sclera, particularly in the posterior part, with a reduction in visual acuity in both eyes. At the same time, the patient was diagnosed with relapsing polychondritis with cardiac involvement, including intermittent second-degree atrioventricular block and paroxysmal atrial fibrillation. Initial treatment with pulse methylprednisolone rapidly improved visual acuity and resolved the scleral inflammation, also the arrhythmias subsided. Following clinical stabilization, long-term oral corticosteroids and cyclophosphamide were introduced. Genetic analysis confirmed the presence of a rare somatic mutation in the UBA1 gene, characteristic of VEXAS syndrome. Bone marrow biopsy and flow cytometry were also performed. After a multidisciplinary consultation with hematologists, treatment with baricitinib, a Janus kinase inhibitor, was initiated. After 12 months of intensive treatment, scleral inflammation is in remission, with restored visual acuity, and the patient's general condition remains stable.
This case emphasizes the critical importance of a multidisciplinary approach in managing patients with rare ocular inflammations, such as bilateral pan scleritis, presenting simultaneously with systemic conditions like relapsing polychondritis and cardiovascular complications. Collaboration between rheumatologists, cardiologists, and ophthalmologists is essential for early diagnosis, effective immunosuppressive therapy, and the prevention of irreversible organ damage and vision loss.

Bilateralni panskleritis pri bolniku z relapsirajočim polihondritisom in VEXAS sindromom
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Namen predstavitve je prikazati primer bolnika z obojestranskim pan skleritisom, relapsirajočim polihondritisom in motnjami srčnega ritma, pri katerem smo ugotavljali prisotnost redke somatske mutacije v genu UBA1 in na podlagi klinične slike in opravljenih preiskav zaključili, da bolnik ustreza merilom za VEXAS sindrom. Kratica VEXAS pomeni V: vakuole, ki so pogosto vidne v celicah pri biopsijah kostnega mozga, E: E1-ubikvitin aktivirajoči encim, kodiran z genom UBA1, X: Gen UBA1 se nahaja na X kromosomu, A: avtoimuna vnetja, S: somatske in ne podedovane mutacije.
55-letni moški je bil napoten v očesno ambulanto zaradi stopnjujočih se težav s pordelimi in bolečimi očmi in zamegljenim vidom obojestransko. S kliničnim pregledom in ultrazvočno preiskavo smo ugotavljali intenzivno vnetje celotne beločnice, zlasti v posteriorni skleri, s padcem vidne ostrine na obeh očeh. Sočasno je bil pri bolniku potrjen relapsirajoči polihondritis s prizadetostjo srca v smislu intermitentnega atrioventrikularnega bloka II. stopnje in paroksizmalne atrijske fibrilacije. Bolnik je bil najprej zdravljen s pulzi metilprednizolona, kar je hitro izboljšalo vid in umirilo vnetje na obeh očesih, tudi motnje ritma so izzvenele. Ob izboljšanju celotne klinične slike je bila uvedena dolgotrajna per os kortikosteroidna terapija skupaj s ciklofosfamidom. Genetska analiza je potrdila prisotnost redke somatske mutacije v genu UBA1, ki je značilna za VEXAS sindrom. Pri bolniku je bila opravljena tudi biopsija kostnega mozga in pretočna citometrija. Po dodatni konzultaciji s hematologi je bilo na podlagi klinične slike in izvidov uvedeno zdravljenje z baricitinibom, zaviralcem janus kinaze. Po 12 mesecih intenzivnega zdravljenja je vnetje sklere v remisiji, vidna ostrina normalna, splošno stanje bolnika pa je stabilno.
S predstavitvijo primera želimo poudariti pomen multidisciplinarnega pristopa pri obravnavi bolnika z redkimi očesnimi vnetji, kot je bilateralni pan skleritis, ob sočasnem pojavu zunaj očesnih težav kot je relapsirajoči polihondritis in kardiovaskularnih zapletih. Sodelovanje med revmatologi, kardiologi in oftalmologi je ključno za pravočasno diagnozo, optimalno imunosupresivno zdravljenje ter preprečevanje trajnih organskih poškodb in izgube vida.