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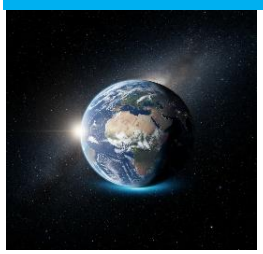

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Relapsing Polychondritis: Clinical Presentation, Diagnosis and Treatment Approaches

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Abstract

Background: Relapsing polychondritis (RP) is a rare, chronic inflammatory disease with a relapsing course that leads to damage of cartilage and other proteoglycan-rich tissues. The variable multisystem presentation and the lack of specific laboratory markers may delay diagnosis, which can result in irreversible deformities and organ-related complications.

Aim: The aim of this paper was to summarise current evidence on the epidemiology, clinical presentation, diagnosis, monitoring, and treatment of RP, with attention to disease heterogeneity and emerging pathogenetic concepts.

Materials and methods: This paper was prepared as a narrative literature review. Searches were conducted in PubMed and Scopus, including review articles, observational studies and clinically relevant case series.

Results: RP most commonly affects the auricles, nose, and laryngo-tracheo-bronchial structures, but may also involve the eyes, inner ear, and cardiovascular system. Diagnosis is primarily clinical, supported by ancillary investigations and exclusion of mimicking conditions; in practice, classification criteria described in the literature are often used. Disease monitoring should include assessment of activity, irreversible damage, and patient functioning; a disease-specific activity index (e.g. RPDAI and RPDAM) may support follow-up. Treatment is individualised and depends on severity and organ involvement; systemic glucocorticoids are the cornerstone, conventional immunosuppressants are used as steroid-sparing therapy, and biologic therapies are considered in refractory cases.

Conclusions: RP remains a major diagnostic and therapeutic challenge due to its rarity, heterogeneous phenotype, and limited availability of high-quality clinical evidence. Early recognition, assessment of dominant organ involvement, regular monitoring, and multidisciplinary care are essential.

Keywords: relapsing polychondritis, chondritis, immunosuppression, glucocorticoids, biologic therapy.

1. Introduction

Relapsing polychondritis (RP) is a rare systemic disease with a chronic, relapsing course, in which the dominant clinical mechanism is inflammation and progressive damage of cartilaginous structures and other proteoglycan-rich tissues. [2,3] The disease may involve multiple sites, typically affecting the auricles, nose, and laryngo-tracheo-bronchial structures; ocular, inner ear, and cardiovascular involvement has also been described, which gives RP a multisystem character. [1,22]

Although RP is classified as a rare disease, it is clinically important due to the risk of irreversible deformities (e.g., of the auricles or nose) and potentially life-threatening complications. [2,3] Airway involvement is considered particularly dangerous, as it may lead to stenosis, airway instability, and respiratory failure. [15,16] Consequently, RP requires high diagnostic vigilance and multidisciplinary management, often involving rheumatology, otolaryngology, pulmonology, cardiology, and ophthalmology. [3]

Diagnosis is difficult for several reasons. First, there is no single laboratory test with high specificity, and inflammatory markers (e.g., CRP and ESR) are non-specific. [10] Second, the clinical picture is heterogeneous and may evolve over time—some patients initially present with articular, ocular, or constitutional symptoms before overt cartilaginous manifestations occur. [2,3] Third, numerous disorders can mimic RP early in the course (e.g., infections, granulomatous diseases, systemic vasculitides), which may further prolong the diagnostic process. [1,12]

In recent years, RP has also been discussed as a biologically heterogeneous entity, and in some patients the phenotype may overlap with autoinflammatory mechanisms and clonal myeloid disorders (e.g., UBA1 variants and VEXAS syndrome). [5,6] These observations expand the current understanding of RP pathogenesis; however, routine clinical practice remains centred on clinical diagnosis and management aimed at controlling inflammation and preventing organ complications. [2,3]

2. Research materials and methods

This paper was prepared as a narrative literature review. Searches were performed in PubMed and Scopus. Publications from 2010 to 2025 were considered. Example keywords (used alone and in combinations) included: relapsing polychondritis, diagnosis, treatment, glucocorticoids,

immunosuppression, biologic therapy, airway involvement, RPDAI, damage, VEXAS, and MAGIC syndrome.

Review papers, observational studies, recommendations/state-of-the-art publications, and clinically relevant case series were included; single case reports were used only as supportive material.

3. Research results

This section summarises the key findings of the narrative review and synthesises current evidence on relapsing polychondritis. The results are presented thematically, covering epidemiology, pathogenesis, clinical presentation, diagnostic approach and criteria, treatment options, disease monitoring, and differential diagnosis.

3.1 Epidemiology

RP is a rare disease, and epidemiological estimates vary depending on the population and methodology. [15,16] In a UK population-based study using the Clinical Practice Research Datalink (1990–2012), the annual incidence was estimated at 0.71 (95% CI 0.55–0.91) per million. [16] Other reviews suggest that the incidence may be higher, reaching up to 3.5 cases per million per year. [17] A Colombian analysis focused on prevalence and reported 11.51 per 100,000 inhabitants (115.1 per million). [18]

Demographically, RP most commonly presents in the 5th decade of life. [15,16] In the UK cohort, the mean age at diagnosis was 55 years in men and 51 years in women, with a median diagnostic delay of 1.9 years. [16] Similar observations were reported in Colombia, where the peak prevalence occurred in the 50–59 age group. [18] A Hungarian study also suggested that a typical patient is middle-aged (45–55 years). [15] Although RP mainly affects adults, paediatric-onset disease can occur and accounts for fewer than 5% of reported cases. [1,4] In children, diagnosis may be delayed or missed because RP is rare and its clinical presentation is heterogeneous. [25]

Sex distribution is inconsistent, indicating a lack of agreement between studies. In the Colombian analysis, women predominated. [18] In contrast, a nationwide Hungarian analysis found a more balanced sex distribution. [15]

Prognosis is closely linked to involvement of critical organs, particularly the respiratory and cardiovascular systems. [16] The UK analysis found that mortality in RP was more than twice

that of the general population, with leading causes of death including respiratory disease, cardiac disease, and malignancies. [16] In the Hungarian study, overall survival was estimated at 83.6–92.9% at 5 years and 75.0–88.3% at 10 years. [15]

3.2 Pathogenesis

The aetiopathogenesis of RP is not fully understood, but multiple findings support immune-mediated cartilage damage. [1,2] In affected tissues, inflammatory infiltrates and secondary cartilage destruction are observed, which clinically results in pain, swelling, and deformity. [1,2] RP is associated with other immune-mediated diseases, which supports the hypothesis of shared mechanisms of inflammatory dysregulation. [1,2] In the nationwide Hungarian cohort, associations with other autoimmune disorders were high (56%). [15]

In recent years, a subgroup of patients with an RP phenotype and pathogenic UBA1 variants has been described, linking some cases to VEXAS syndrome and suggesting contributions from autoinflammatory pathways and clonal haematological disturbances. [5,6]

3.3 Clinical presentation

Auricular chondritis

Auricular cartilage inflammation is among the most common manifestations of RP. [1,2] Examination typically shows sudden pain, erythema, swelling, and warmth of the cartilaginous part of the ear, with characteristic sparing of the ear lobe (which lacks cartilage). [1,2] Recurrent episodes may lead to laxity and deformity of the auricle, sometimes resembling a “cauliflower ear”. [20]

Nasal chondritis

Nasal involvement may present with pain and tenderness over the nasal bridge and swelling of surrounding soft tissues. [1] Chronic destruction of the septal and dorsal cartilage can result in a saddle-nose deformity. [2,3]

Laryngo-tracheo-bronchial involvement

Airway cartilage involvement may cause hoarseness, cough, dyspnoea, stridor, and recurrent respiratory infections. [1,2] In severe cases, progressive stenosis and airway instability can develop and may be directly life-threatening. [2,3] In selected situations, interventional procedures (e.g., tracheotomy, airway stenting) may be required, especially in progressive obstruction. [1,3]

Arthritis

In RP, arthritis is usually non-erosive and non-deforming and may present as recurrent, often asymmetric oligoarthritis. [1,2] Small joints of the hands and larger joints (including knees) are commonly involved, and the pattern may be migratory. [1,2] In paediatric patients, the knees and ankles are commonly affected. [12]

Ocular involvement

Ocular manifestations (e.g., conjunctivitis, episcleritis, and scleritis) are frequent extra-cartilaginous features. [1,2] Episcleritis and scleritis are reported among the most common ocular presentations, while more severe complications (e.g., uveitis) may occur. [1,3] Severe scleritis may threaten vision and requires prompt systemic treatment and close ophthalmological cooperation. [2,3]

Inner ear

Involvement of the auditory and vestibular system may present with conductive and/or sensorineural hearing loss, tinnitus, and episodic vertigo. [1,22] Conductive hearing loss may result from narrowing of the external auditory canal, Eustachian tube dysfunction, or otitis media. [1] Sensorineural hearing loss may relate to inner-ear involvement and in some cases may progress to deafness. [22]

Skin

Cutaneous manifestations are usually non-specific (e.g., purpura, nodules, livedo, ulcers, aphthae) and may be more frequent in patients with systemic disease or haematological disorders. [19, 21]

Cardiovascular system

A minority of patients develop cardiovascular complications, including valvular involvement (mitral or aortic). [22,29] Myocarditis, pericarditis, myocardial infarction, and inflammatory involvement of vessel walls (including aortic aneurysms) have been reported. [29] Because cardiovascular involvement may be oligosymptomatic, periodic cardiological assessment is recommended when involvement is suspected. [2,3]

Renal involvement

Renal involvement is a rare manifestation, but it is associated with poorer prognosis and increased mortality. [1,2] Contemporary reviews suggest that the frequency may be lower than

historically reported, partly due to misclassification in patients with granulomatosis with polyangiitis (GPA), which can mimic RP with overlapping organ involvement. [2]

Neurological involvement

Neurological manifestations are uncommon (around 3%) and most often involve cranial nerves, particularly V and VII. [1] Reported features include headaches, aseptic meningitis, encephalitis, ataxia, seizures, and psychiatric symptoms. [1]

Phenotypic groups

RP demonstrates marked clinical heterogeneity, and distinct phenotypes with differing dominant organ involvement and prognosis have been described. [19] Based on the analysis of a French patient cohort, three major clinical phenotypes can be distinguished. [24]

- **Mild phenotype** - the most common group, typically dominated by auricular and nasal chondritis and associated with the most favourable prognosis [19, 24]
- **Respiratory-predominant phenotype** - a less frequent presentation characterised mainly by involvement of laryngeal and tracheal cartilage, reflecting predominant airway disease [19, 24]
- **Haematological/VEXAS-associated phenotype** - the least common but prognostically most severe group, more often linked to haematological disorders and VEXAS syndrome and associated with a higher burden of serious cardiovascular complications [19, 24]

3.4 Diagnostic tests

There is no single laboratory marker specific for RP. [2,3] During flares, elevated inflammatory markers (e.g., CRP and ESR) are common but non-specific and cannot be used as a stand-alone diagnostic basis. [1,3] Autoantibodies (e.g., ANA, RF, ANCA) may be useful for identifying coexisting conditions and for differential diagnosis but are not diagnostic for RP. [1,2]

Cartilage biopsy may support diagnosis in equivocal cases by showing inflammatory changes and cartilage destruction. However, histopathological findings are not pathognomonic. [32] Histopathology may also show leukocytoclastic vasculitis, vascular thrombosis, and neutrophilic infiltrates, but these features are nonspecific. [31]

Imaging is tailored to the dominant phenotype, particularly in suspected airway or cardiovascular involvement. [2,3]

CT commonly used to evaluate airway wall abnormalities, narrowing, and cartilage-related changes. [27] A key limitation is that CT may not reliably distinguish active inflammation from chronic fibrotic remodeling. [27]

MRI may show cartilage and perichondrial inflammation with contrast enhancement, potentially even in earlier stages of disease. [25]

3.5 Diagnostic criteria

Diagnosis of RP is primarily clinical, supported by ancillary testing and exclusion of other conditions. [2,3] In practice, the following classification criteria are commonly referenced:

- **McAdam criteria (1976):** RP is diagnosed when at least three of six features are present: (1) auricular chondritis, (2) non-erosive inflammatory polyarthritis, (3) nasal chondritis, (4) ocular inflammation, (5) respiratory tract chondritis, (6) hearing loss or vestibular dysfunction; histological confirmation is not required. [7]
- **Damiani and Levine criteria (1979):** RP can be diagnosed with (a) one McAdam feature plus histological confirmation, or (b) two McAdam features plus a favourable response to glucocorticoids or dapsone. [8]
- **Michet criteria (1986):** RP is diagnosed with proven inflammation in at least two of three sites (auricular, nasal, laryngo-tracheal), or proven inflammation in one of these sites plus at least two “minor” features (hearing loss, ocular inflammation, vestibular dysfunction, seronegative arthritis). [9]

3.6 Treatment

There are no universally accepted RP treatment guidelines based on randomised clinical trials; management relies mainly on reviews, case series, and expert experience. [2,3] Treatment is individualised according to severity and organ involvement, aiming for rapid control of inflammation and prevention of irreversible damage and life-threatening complications. [2,3]

Mild and moderate disease

In mild disease (e.g., limited to the ear, nose, or joints), anti-inflammatory agents are used, and if response is insufficient, low-dose glucocorticoids may be considered. [30] In selected cases, dapsone or colchicine has been described, especially for recurrent inflammatory symptoms and skin manifestations; decisions should be individualised. [3,13]

Glucocorticoids

Systemic glucocorticoids are the cornerstone for moderate-to-severe RP, especially with ocular, airway, cardiovascular, or central nervous system involvement. [2,3] In severe flares or life-threatening involvement, intravenous methylprednisolone pulses are commonly used in clinical practice. [2,3] Long-term steroid therapy carries a substantial risk of iatrogenic complications, which supports the use of steroid-sparing strategies. [2,3]

Conventional immunosuppressants (csDMARDs)

Immunosuppressive agents are used as steroid-sparing therapy or in disease refractory to glucocorticoid monotherapy. [2,3] Agents used in practice include methotrexate and azathioprine, and cyclophosphamide has been considered in severe organ-threatening disease. [2,3] Selection should consider the organ phenotype, comorbidities, and safety profile. [2,3]

Biologic therapy

Biologic therapy is considered in RP refractory to conventional treatment, although evidence mainly comes from case series and reviews. [2,13] Reported biologic therapies include TNF inhibitors, tocilizumab (anti-IL-6), anakinra (anti-IL-1), and abatacept; responses may be variable and require individual assessment. [13,14] Biologic therapy should ideally be initiated in experienced centres with a plan for monitoring adverse effects and infections. [2,3]

Organ-directed and interventional management

With airway involvement—especially progressive obstruction—interventional procedures (e.g., tracheotomy, airway stenting) may be required alongside immunosuppression. [1,3] Severe ocular disease (e.g., severe scleritis) requires aggressive systemic therapy and close cooperation with ophthalmology. [2,3] Cardiac and aortic involvement requires rheumatology-internal medicine management with cardiology input, and in selected cases cardiothoracic or vascular interventions. [2,3]

3.7 Disease monitoring

Monitoring should include assessment of disease activity, irreversible damage, and patient functioning. [2,3] RPDAI (Relapsing Polychondritis Disease Activity Index) is a a disease-specific activity index for assessing RP activity over a defined time period and is used in research and clinical follow-up. [10,11] RPDAM (Relapsing Polychondritis Damage

Assessment Measure) has been described as a tool for evaluating irreversible disease consequences relevant for long-term care. [10,26]

Laboratory parameters (CRP, ESR) may support assessment of non-specific inflammatory activity and treatment response but are not specific to RP and should be interpreted in clinical context. [1,3] The scope of follow-up testing should depend on the dominant organ manifestations (e.g., ENT assessment and airway evaluation in respiratory symptoms; ophthalmological evaluation in ocular involvement). [2,3] Cardiological monitoring (including echocardiography) is particularly important when cardiac/aortic involvement is suspected. [2,29]

RP significantly impacts quality of life, which supports incorporating patient-reported burden and functional symptoms into long-term follow-up. [12] Special vigilance is warranted in patients with early tracheal involvement, which has been associated with a higher risk of recurrence. [23]

3.8 Differential diagnosis

Auricular involvement should be differentiated from bacterial infections, trauma, and other causes of external ear inflammation. [1,2]

Nasal deformities and chronic upper airway inflammation require differentiation from granulomatous diseases—especially granulomatosis with polyangitis—as well as infectious conditions (leprosy, congenital syphilis) and vasculitides. [1,25]

Arthritis should be differentiated from rheumatoid arthritis and infectious causes. [1,2]

Ocular inflammation seen in patients evaluated for RP can also be caused by infections or by systemic inflammatory diseases such as juvenile idiopathic arthritis, granulomatosis with polyangiitis, polyarteritis nodosa, sarcoidosis, Behçet’s disease, systemic lupus erythematosus, and seronegative spondyloarthropathies, so these entities should be considered in the differential diagnosis. [25]

Differential diagnosis and phenotyping should also consider RP overlapping with VEXAS syndrome (UBA1), particularly in patients with haematological features. [5,6] The MAGIC syndrome (Mouth And Genital ulcers with Inflamed Cartilage) has also been described as an overlap between RP and Behçet’s disease. [28]

4. Discussion

RP remains a rare but clinically important disease because its chronic, relapsing course can lead to irreversible organ damage. [2,3] The heterogeneous clinical spectrum—from mild auricular/nasal disease to severe respiratory or cardiovascular involvement—means that diagnostic and therapeutic strategies must be individualised and based on the dominant organ phenotype. [2,3] Phenotype stratification is relevant not only descriptively but also prognostically and organisationally (specialist consultations, follow-up schedules, intensity of immunosuppression). [19]

Population data indicate that mortality in RP may be significantly higher than in the general population, with respiratory and cardiac disease among leading causes of death. [16] At the same time, European cohort data suggest that long-term survival can be relatively good. [15]

Recent data also suggest that some initial manifestations may predict a higher recurrence risk. [23] In particular, a study evaluating recurrence risk factors found that tracheal involvement at initial presentation was independently associated with an increased risk of relapse (HR 4.266). [23] This supports proactive monitoring of patients with early airway involvement. [2,23]

Diagnostic difficulty arises from the absence of a single highly specific test, the non-specific nature of inflammatory markers, and changing phenotype over time. [2,20] In this context, clinical criteria (McAdam; Damiani and Levine; Michet) remain practical foundations for diagnosis, although early disease may not meet full criteria. [7,8] Broad differential diagnosis remains essential because several conditions can mimic RP and misclassification may affect treatment and assessment of complications (e.g., renal involvement). [1,2]

The identification of patients with RP-like phenotypes carrying pathogenic UBA1 variants has shifted understanding of a subset of cases toward VEXAS spectrum disease. [5,6] In selected patients—especially those with haematological abnormalities or atypical, severe courses—additional evaluation for VEXAS may be justified and may influence prognosis and treatment decisions. [5,6]

Despite advances in phenotype description and therapeutic options, there is still a lack of universal recommendations supported by high-quality randomised clinical trials, and most therapeutic decisions are derived from reviews, case series, and centre experience. [2,3] In practice, glucocorticoids and conventional immunosuppressants (e.g., methotrexate,

azathioprine; cyclophosphamide in severe organ involvement) remain central, while biologic therapies are mainly considered in refractory disease with limited evidence and variable responses. [2,13] Implementation of activity and damage measures (RPDAI, RPDAM) may standardize assessment, improve disease management and support long-term treatment adjustment. [10,11]

5. Conclusions

Relapsing polychondritis (RP) is a rare multisystem disease with a relapsing course, and in clinical practice the diagnosis remains primarily based on the clinical picture due to the lack of a single highly specific test. [2,3] Clinically, early identification of dominant organ involvement and prompt initiation of inflammation-controlling management are essential to reduce the risk of irreversible damage and complications. [2,3] Prognosis depends largely on critical organ involvement, particularly the respiratory and cardiovascular systems, which supports multidisciplinary care and regular monitoring. [2]

Glucocorticoids and immunosuppressants remain the backbone of therapy, while biologic therapies are mainly considered in refractory cases and evidence quality remains limited. [2,13] Long-term care should combine assessment of activity and damage supported by tools such as RPDAI and RPDAM. [10,11]

In selected patients with atypical, severe disease and/or haematological features, extending the diagnostic work-up toward VEXAS/UBA1 may be appropriate and can influence subsequent clinical decisions. [5,6]

Disclosure

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