EBMT Registry

Autoimmune disorders

Guide to the completion v2.2 of the EBMT data collection form:

Autoimmune_Disorders_v2.2

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EBMT Registry

EBMT Clinical Research & Registry Department

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Table of Contents

Introduction

Please make sure you have already checked the **Introduction to the EBMT Registry Completion Guidelines** document latest version available under *Manuals and Reference Documents* section on EBMT website.

Autoimmune disorders

Autoimmune disorders (ADs) represent a heterogeneous group of diseases that affect approximately 5–10% of the population in industrialised countries with increasing incidence. Although the clinical phenotypes of ADs vary widely depending on the type of tissue and immunologic components involved, their common characteristic is a break of self-tolerance with chronic inflammation of target organs or multiple organ systems. It is widely accepted that autoimmunity develops in a genetically predisposed population, where a combination of certain epigenetic factors and environmental triggers may result in the activation of normally quiescent autoreactive cells that escape self-regulation.

This Autoimmune Disorders diagnosis form must be completed only if an AD is the indication for HCT/CT. If it is concomitant disease or secondary disease, please complete a **non-indication diagnosis form**.

If VEXAS syndrome concurrent with MDS, VEXAS syndrome should be reported as non-indication diagnosis using the **Non-indication diagnosis form.**

No data items should be left blank unless specifically stated in the definition.

Disease

Date of diagnosis

Report the date of the first diagnosis of the disease. Add the date indicated by a physician within the patient's medical record.

Classification

Select the subclassification that is appropriate for the AD and check the box next to it.

Connective tissue

Select one of the options from the list:

Systemic sclerosis (scleroderma) is a heterogeneous condition of unknown aetiology characterised by microvascular injury and the deposition of excess collagen in the skin and internal organs.

Two clinical patterns of lung involvement are recognized: precapillary vascular disease and interstitial lung disease (ILD), which has become the leading cause of death in SSc since the introduction of ACE-inhibitors to treat previously fatal renal crises. Vasculopathy may lead to pulmonary arterial hypertension (PAH) even in the absence of significant fibrosis. PAH, defined as a mean pulmonary artery pressure higher than 25 mmHg at rest or higher than 30 mmHg during exercise in the absence of left-sided heart disease, occurs in at least 10% of SSc patients and is associated with high mortality. Lung fibrosis has been found in approximately 70% of SSc patients at autopsy. Most studies differentiate scleroderma-associated PAH and ILD as 2 separate pathological processes, concentrating on one or the other. Many patients though have both conditions. ILD is more frequent in dcSSc (53%) than in lcSSc (35%), whereas PAH is diagnosed in a similar frequency within the two subsets.

Skin thickening and Raynaud's phenomenon are the key clinical features of systemic sclerosis. For scientific purposes, classification criteria have been updated by joint effort of the American College of Rheumatology (ACR) and European Alliance of Associations for Rheumatology (EULAR) in 2013, referred to as 2013 ACR/EULAR criteria (1). Patients with a total score of 9 are classified as having definite SSc.

SSc type

A further subclassification can be made in diffuse cutaneous SSc, limited cutaneous SSc, SSc sine scleroderma. Select the SS type from the list:

- **Diffuse cutaneous SSc** is characterised by skin involvement on the upper arms and trunk, is associated with an early incidence of interstitial lung disease, hypertensive crisis and renal failure, diffuse gastrointestinal disease, and myocardial involvement, and the presence of anti-topoisomerase (or anti-Scl70) antibodies.
- **Limited cutaneous SSc** is characterised by skin involvement limited to hands, feet, face, and/or forearms, and is associated with a high incidence of anticentromere autoantibodies (ACA) (70-80%), the existence for years of Raynaud's phenomenon and a significant late incidence of pulmonary hypertension. The acronym CREST (Calcinosis, Raynaud's phenomenon, Esophageal dysmotility, Sclerodactyly, and Telangiectasia) fits into this subclassification.
- **SSc sine scleroderma** is characterised by Raynaud's phenomenon (RP), visceral involvement without thickening of the skin, and anticentromere antibodies (ACA).

If the SSc type is not available in the list, check the **Other** box and report the type in the textbox in English.

Systemic lupus erythematosus (SLE), currently classified according to the 2019 EULAR/ACR criteria (1), is the most frequent connective tissue disease with a prevalence of 40 to 50 per 100 000 people. Its exact origin is still unknown, but heredity, environment, and hormonal changes may be involved. SLE is a

heterogeneous chronic multisystemic autoimmune inflammatory disorder, with various types of clinical symptoms affecting mostly females (> 85%), with a higher frequency in certain ethnic groups, especially among black people.

Classification criteria:

In 2019, the classification criteria for SLE have been updated by EULAR/ACR initiative, achieving a sensitivity of 96.1% and specificity of 93.4%, compared to 96.7% sensitivity and 83.7% specificity of the Systemic Lupus International Collaborating Clinics (SLICC) 2012 criteria (2). The 2019 EULAR/ACR classification criteria for SLE include positive ANA at least once as obligatory entry criterion; followed by additive weighted criteria grouped in 7 clinical (constitutional, hematologic, neuropsychiatric, mucocutaneous, serosal, musculoskeletal, renal) and 3 immunological (antiphospholipid antibodies, complement proteins, SLE-specific antibodies) domains, and weighted from 2 to 10. Patients accumulating ≥10 points are classified.

Mixed connective tissue disease (MCTD) is a rare systemic autoimmune disease with an overlapping feature of at least two connective tissue diseases (CTD), including systemic lupus erythematosus (SLE), systemic sclerosis (SSc), polymyositis (PM), dermatomyositis (DM) and rheumatoid arthritis (RA) along with the presence of a distinctive antibody, anti-U1-ribonucleoprotein (RNP) previously known as an antibody to extractable nuclear antigen (ENA). MCTD has no unique clinical features, and there is a considerable inter-individual variation in clinical manifestations. Alarcon-Segovia is one of the regularly used diagnostic criteria that consists of a high titer of positive anti-U1-RNP (over 1 per 1600) and three or more of the following clinical manifestations: Raynaud phenomenon, hand edema, synovitis, histologically proven myositis, and acrosclerosis.

A revised set of diagnostic criteria for MCTD, which divides the features of the disease into the following four categories, was proposed by a consensus panel in Japan in 2019 (2,3). These include:

- Raynaud phenomenon
- Immunologic manifestation such as anti–U1-RNP antibody
- Organ involvement includes pulmonary arterial hypertension, aseptic meningitis, trigeminal neuropathy

Overlapping manifestations of SLE-like, systemic sclerosis, polymyositis, and dermatomyositis may be present. MCTD may be diagnosed based upon a thorough clinical evaluation, a detailed patient history, identification of characteristic findings and specialized tests (i.e. antibody mentioned above).

Polymyositis/Dermatomyositis (PM/DM) are different disease subtypes of idiopathic inflammatory myopathies (IIMs). The main clinical features of PM and DM include progressive symmetric, predominantly proximal muscle weakness. Laboratory findings include elevated creatine kinase (CK), autoantibodies in serum, and inflammatory infiltrates in muscle biopsy. Dermatomyositis can also involve a characteristic skin rash. Both polymyositis and dermatomyositis can present with extra muscular involvement.

In order to make a diagnosis of PM/DM, the criteria developed during the ENMC international workshop in 2003 are used (4).

Sjögren syndrome is a chronic autoimmune disorder characterised by inflammatory destruction of the body's exocrine glands and may be considered primary or secondary, in association with other autoimmune disorders, such as collagen vascular diseases. In order to make a diagnosis of Sjögren's syndrome, the American-European Consensus Criteria are used (5).

Antiphospholipid syndrome is characterised by thrombotic and/or pregnancy morbidity associated with the presence of persistent antiphospholipid antibodies (aPLs).

The updated international consensus (Sydney) classification (ICS) criteria for definite antiphospholipid syndrome require the presence of a lupus anticoagulant (LA) and/or IgG or IgM anticardiolipin antibodies (aCL) present in medium or high titre (i.e. >40 GPL or MPL or >99th percentile), and/or anti-β2glycoprotein-1 (aβ2GPI) (IgG and/or IgM) >99th percentile (5)(6). These aPL should be persistent, defined as being present on two or more consecutive occasions at least 12 weeks apart.

If the connective tissue disease is not available in the list, select **Other connective tissue disease** and specify the disease name in the textbox in English.

Vasculitis

Granulomatosis with polyangiitis (GPA); formerly Wegener granulomatosis, is characterised by necrotizing granulomatous inflammation involving the ears, nose, and upper and lower respiratory tracts, and necrotizing vasculitis affecting predominantly small- to medium-sized vessels, often including necrotizing glomerulonephritis. ACR/EULAR-endorsed classification criteria are used for GPA diagnosis (7).

Classical polyarteritis nodosa is a systemic vasculitis characterised by necrotizing inflammatory lesions that affect medium-sized and small muscular arteries, preferentially at vessel bifurcations. These lesions result in microaneurysm formation, aneurysmal rupture with haemorrhage, thrombosis, and, consequently, organ ischemia or infarction.

Microscopic polyarteritis nodosa is a systemic necrotizing vasculitis without immune globulin deposition (pauci-immune) that affects mainly small vessels. It may begin as a pulmonary-renal syndrome with

rapidly progressive glomerulonephritis and alveolar haemorrhage, but the pattern of the disease depends on the organs affected.

Eosinophilic granulomatosis with polyangiitis (EGPA); formerly Churg-Strauss is a form of vasculitis that is histologically defined by eosinophil-rich, necrotizing granulomatous inflammation primarily involving the respiratory tract, along with necrotizing vasculitis of small- to medium-sized arteries. EGPA is considered a form of antineutrophil cytoplasmic antibody (ANCA)–associated vasculitis (AAV), along with granulomatosis with polyangiitis (GPA) and microscopic polyangiitis (MPA). ANCAs are detected in ~40–60% of patients with EGPA and are typically directed against myeloperoxidase (MPO). ACR/EULAR-endorsed classification criteria are used for EGPA diagnosis (7,8).

Behçet syndrome is an inflammatory vasculopathy with multisystemic involvement. The clinical course usually follows a relapsing-remitting course with heterogeneous clinical manifestations. There are no pathognomonic laboratory tests to diagnose BD, and as such, the diagnosis is based on clinical criteria (7–9).

Takayasu arteritis is one of the major forms of large-vessel vasculitis (LVV). TAK is a chronic disease defined by granulomatous inflammation affecting the aorta and its primary branches. Complications from vascular damage can result in substantial morbidity including stroke, myocardial infarction, mesenteric ischaemia, and limb claudication. ACR/EULAR-endorsed classification criteria are used for TAK diagnosis (10).

If the vasculitis sub-class is not available in the list, select **Other** and specify the disease name in the textbox in English.

Arthritis

Select one of the options from the list:

Adult onset stills disease (AOSD) is a rare, idiopathic, inflammatory disorder characterised by arthralgia, evanescent, salmon-coloured rash, and quotidian or double quotidian fevers. Lymphadenopathy, pharyngitis, splenomegaly, myalgias, and serositis are also commonly seen with this disease. Frequently seen laboratory abnormalities include leukocytosis, transaminitis, elevated ferritin levels, increased acute phase reactant concentrations, and aberrant production of proinflammatory cytokines. While the inciting aetiology of this syndrome is unknown, both viruses and bacteria have been isolated in patients with AOSD leading to the hypothesis that infection triggers an autoimmune response. A number of different diagnostic criteria have been published, with the most sensitive and widely used being the Yamaguchi Criteria (11).

Rheumatoid arthritis is a chronic systemic and autoimmune disease that affects approximately 1% of the world's population. Being characterised mainly by persistent articular inflammation, this condition affects

the synovial membranes of the joints, leading to joint destruction, loss of functions, and osteoarticular disabilities. In the disease's progression, bone and cartilage are destroyed, which brings deformities to the patients. The 2010 ACR/EULAR-endorsed classification criteria are currently used for RA diagnosis (12).

Psoriatic arthritis/psoriasis is inflammatory arthritis with multiple manifestations: peripheral/axial arthritis, enthesitis, dactylitis, psoriasis, and nail involvement.

The diagnosis of psoriatic arthritis (PsA) often is missed, partly because patients may present with inflammatory spinal pain, tendinitis, enthesitis, or dactylitis rather than a "true arthritis." If PsA is not identified early and managed appropriately, progressive joint damage with deformities and disability may result. The CASPAR criteria permit the diagnosis of PsA in spite of low RF positivity. The CASPAR criteria consist of confirmed inflammatory articular disease (joint, spine, or entheseal) with at least 3 points from the following features: current psoriasis (assigned a score of 2 points; all other features are assigned a score of 1), a history of psoriasis or a family history of psoriasis (unless current psoriasis is present), dactylitis, juxta-articular new bone formation (hands or feet), rheumatoid factor (RF) negativity (except latex test), and psoriatic nail dystrophy (13).

Juvenile idiopathic arthritis (JIA) has as a main aspect is the presence of chronic arthritis. Arthritis is defined as joint swelling and/or pain on movement or palpation of the joint. The duration of arthritis must be at least 12 weeks. Known causes of joint swelling such as infections, malignancies or metabolic diseases must be excluded. The ILAR (International League Against Rheumatism) subclassification of Juvenile Idiopathic Arthritis (JIA) is nowadays the most commonly used (14). Of these, the polyarticular forms (with or without rheumatoid factor) and the systemic onset forms are the most severe subtypes.

Based on the ILAR subclassification, there are two main subclasses of JIA:

- **Juvenile idiopathic arthritis (JIA), systemic (Still's disease)** which is defined as arthritis in 1 or more joints with or preceded by daily spiking fever of at least 2 weeks duration and accompanied by 1 or more of the following: Pink evanescent rash, generalised lymph node enlargement, hepatosplenomegaly or serositis.
- **Juvenile idiopathic arthritis (JIA), articular**

Juvenile idiopathic arthritis (JIA), articular

Depending on the manifestations at onset of Juvenile idiopathic arthritis (JIA), articular can be subdivided into:

- **oligoarticular onset** - arthritis affecting 1 to 4 joints during the first 6 months of the disease.
- **polyarticular onset** - arthritis affecting 5 or more joints during the first 6 months of the disease.

If the juvenile idiopathic arthritis sub-class is not available in the list, select Other juvenile idiopathic arthritis and specify the disease name in the textbox in English.

Examples of other JIA sub-classes are:

- **Psoriatic JIA:** arthritis and psoriasis and at least 2 of the following: dactylitis, nail pitting, and psoriasis in a first-degree relative.
- **Enthesitis-related JIA:** arthritis and enthesitis and at least 2 of the following: sacroiliitis, presence of HLA-B27, onset in males over 6 years of age, presence of acute anterior uveitis,
- **Unclassified JIA:** arthritis that fulfils criteria in no category or in 2 or more of the other categories

If the arthritis sub-class is not available in the list, select Other and specify the disease name in the textbox in English.

Neurological diseases

Select one of the options from the list:

Multiple sclerosis is a chronic, inflammatory, demyelinating disease of the central nervous system, and is one of the most common causes of neurological disability in young adults.

Different criteria are used for assigning the MS diagnosis. In practice, multiple sclerosis is classified into clinical subtypes on the basis of the patient's clinical course: relapsing-remitting (acute attacks followed by recovery), primary progressive (gradual worsening from onset), and secondary progressive (relapsing-remitting at onset but gradual worsening later in the disease course) (15). In the 2013 revision, clinical and radiological disease activity and progression were introduced as modifiers of the basic clinical courses to better reflect treatment-relevant aspects of the disease, such as relapses (16). The revised 2017 McDonald's criteria allow for earlier diagnosis in the setting of a single clinical attack and corresponding MRI findings of symptomatic or asymptomatic, enhancing T1 or non-enhancing T2 lesions typical of MS, and/or presence of cerebrospinal fluid (CSF) specific oligoclonal bands (17).

The historical clinical subtypes include clinically isolated syndrome (CIS), relapsing-remitting MS (RRMS), primary progressive MS (PPMS), and secondary progressive MS (SPMS) (17). A CIS is defined as a first demyelinating episode with features typical of an MS attack such as optic neuritis, brainstem or spinal cord lesion, but not yet fulfilling full criteria for MS. A more recent refinement of MS disease subtype classification proposed by Lublin et al. is similar with the additional caveat of modifying MS subtypes as “active” or “not active” based clinical relapse and/or MRI activity (16,17). The term RMS covers “relapsing-remitting disease (RRMS) and active secondary progressive disease”.

The date of diagnosis, in this case, is the date when the patient would fulfil any criteria (radiologic, laboratory, or clinical) for the diagnosis of MS.

Myasthenia gravis is a neuromuscular transmission disorder characterised by fluctuating weakness and fatigability of voluntary muscles (ocular, bulbar, limbs, neck and respiratory) without loss of reflexes or impairment of sensation or other neurologic function. The neuromuscular transmission defect is usually demonstrated by pharmacological and electrophysiological tests. MG is an autoimmune disorder and is usually (80% of MG patients) mediated by autoantibodies to the acetylcholine receptor (AChR, AChR-MG); in the 20% of patients that are not positive for AChR antibody, up to 50% have antibodies to muscle-specific kinase (MuSK, MuSK-MG). The remaining patients are negative for both antibodies (SNMG), but the evidence is accumulating which strongly indicates that other, as yet unidentified, autoantibodies are responsible for SNMG. The diagnosis is based on clinical appraisal and confirmed by one or more pharmacological, electrophysiological, or serological tests. Imaging studies are essential to search for a thymoma.

Chronic inflammatory demyelinating polyneuropathy (CIDP) is the most common chronic immune-mediated inflammatory polyneuropathy and includes several subtypes that belong to the spectrum of causally treatable neuropathies. Clinically CIDP is classified into 'typical' and 'atypical' cases; typical CIDP is a symmetrical polyneuropathy affecting proximal and distal muscles equally, whereas atypical CIDP includes 'distal acquired demyelinating symmetric' (DADS), and multifocal acquired demyelinating sensory and motor neuropathy (MADSAM, or Lewis-Sumner syndrome [LSS]). DADS is a symmetrical length-dependent sensory or sensorimotor neuropathy, often associated with an IgM paraprotein and markedly increased distal motor latencies. The features are characteristic of demyelinating neuropathy with antimyelin-associated glycoprotein (MAG) antibodies, but in the EFNS/PNS criteria, anti-MAG neuropathy is excluded from CIDP, largely because of the presence of a specific antibody and different treatment response. LSS has a multifocal distribution, and the electrophysiological hallmark of the disease is the presence of a conduction block. In addition, pure motor and sensory CIDP variants have been reported, the latter sometimes restricted to sensory nerve roots (chronic immune sensory polyradiculopathy). A rare chronic ataxic neuropathy associated with ophthalmoplegia, IgM paraprotein, cold agglutinins and disialosyl (ganglioside) antibodies is known by the acronym CANOMAD.

Neuromyelitis optica (NMO) or NMO spectrum disorders (NMOSD) is an autoimmune disease of the central nervous system (CNS) that mainly affects the optic nerves and spinal cord. It is sometimes referred to as NMO spectrum disorder or NMOSD. The disease is caused by abnormal auto-antibodies that bind (attach) to a protein called aquaporin-4, activating other parts of the immune system and causing inflammation and damage to cells. This also results in the loss of myelin in the brain and spinal cord.

NMO is different from multiple sclerosis (MS). Attacks are usually more severe in NMO than in MS, and NMO is treated differently from MS. Most individuals with NMO experience clusters of attacks days to months or years apart, followed by partial recovery during periods of remission.

If the neurological diseases subclass is not available in the list, select Other autoimmune neurological disorder and specify the disease name in the textbox in English.

Haematological diseases

Select one of the options from the list:

Idiopathic thrombocytopenic purpura (ITP) is an autoimmune-mediated haematological disorder affecting platelets. A patient's immune system produces antibodies directed against platelet antigens, resulting in platelet destruction and suppression of platelet production in the bone marrow. Patients with ITP are, therefore, at risk of serious bleeding events.

ITP is defined as isolated thrombocytopenia (platelet count <100 x 10^9 /L) with no associated causes or disorders. Diagnosis of ITP is one of exclusion; there is no standard test for ITP. Diagnosis is usually made based on the patient's medical history, physical examination, complete blood count (CBC), and examination of a peripheral blood smear (18).

Haemolytic anaemia (AIHA) is a decompensated acquired haemolysis caused by the host's immune system acting against its own red cell antigens. Initial simple investigations for haemolytic anaemia, caused by the destruction of red blood cells and increased haemoglobin catabolism, are decreased levels of haemoglobin, and an increase in efforts of bone marrow to regenerate products. One of the main laboratory values that aids in the diagnosis of hemolytic anaemia is an elevated reticulocyte count, as the bone marrow is attempting to produce increased amounts of RBCs. This can be seen in other disease processes such as blood loss anaemia; therefore, one must be cautious in taking a thorough history as well as evaluating other lab values that should also be altered, including LDH, haptoglobin, and indirect bilirubin. Hemolysis can be seen in many rare conditions from paroxysmal nocturnal hemoglobinuria to blood transfusions or mechanical circulatory support; therefore, a wide differential must be ruled out. Once hemolysis is confirmed, further investigation is needed to establish if that hemolysis is immune (AIHA), principally by the direct anti-globulin test (DAT). The standard DAT demonstrates that immunoglobulin G (IgG) and/or complement (usually C3d) is bound to the red cell membrane. Autoantibodies can also be of IgM and IgA classes, and, in some circumstances, an extended DAT panel can be used to detect these.

Evan syndrome is an autoimmune condition that presents with two or more cytopenias, which commonly includes autoimmune haemolytic anaemia (AIHA) and idiopathic thrombocytopenic purpura (ITP), with or without immune neutropenia (only in 15% of cases, according to a report). The type of AIHA that presents in Evans syndrome is warm AIHA, in which IgG antibodies react with red blood cell (RBC) surface antigens

at body temperature, as opposed to cold AIHA. In ITP, the immune system is directed against GPIIb/IIIa on the platelets.

Autoimmune lymphoproliferative syndrome (primary diagnosis, not subsequent to transplant) is a human genetic disorder of lymphocyte apoptosis resulting in an accumulation of lymphocytes, childhood-onset chronic lymphadenopathy, splenomegaly, multilineage cytopenias, and an increased risk of B-cell lymphoma.

There is no single laboratory abnormality that is diagnostic of ALPS. However, select findings are either required for the diagnosis or strongly support it. Per the 2010 revised criteria, patients should demonstrate an elevated number of circulating double-negative α/β T cells comprising greater than 1.5% of total lymphocytes or at least 2.5% of total T cells. In combination with chronic lymphadenopathy or splenomegaly for greater than 6 months, these constitute the 2 required findings for a clinical diagnosis of ALPS and should trigger ancillary testing.

If the haematological diseases subclass is not available in the list, select Other haematological autoimmune disease and specify the disease name in the textbox in English.

Inflammatory bowel diseases (IBD)

Recently new diagnostic consensus criteria for diagnosis of IBD have been jointly developed by the European Crohn's and Colitis Organisation [ECCO] and the European Society of Gastrointestinal and Abdominal Radiology [ESGAR] (19).

Select one of the options from the list:

Celiac disease is an autoimmune condition characterised by a specific serological and histological profile triggered by gluten ingestion in genetically predisposed individuals. An intestinal (duodenal) biopsy is considered the "gold standard" for diagnosis. The World Gastroenterology Organization recommends pathologists use a modified Marsh classification for interpretation (20).

Crohn's disease is an inflammatory bowel disease that is characterised by chronic inflammation of any part of the gastrointestinal tract and has a progressive and destructive course. There is no gold standard for diagnosing Crohn's disease (CD) yet and the diagnosis is based on the comprehensive evaluation of clinical, endoscopic, histological, surgical, imaging, and laboratory findings. The symptoms of CD vary. Primarily, ileocolonoscopy and biopsy are recommended for the diagnosis of CD. The endoscopic findings of CD include longitudinal ulcers, cobblestone-like appearance, discontinuous multiple aphthous ulceration, asymmetrical inflammation, and skipped lesions. In addition, fissures, fistulas, and strictures may be found. The histopathologic findings of CD include non-caseating granulomas and other chronic enteritis findings.

Ulcerative colitis is a chronic inflammatory condition that causes continuous mucosal inflammation of the colon, usually without granulomas on biopsy. It affects the rectum and to a variable extent the colon in a continuous fashion, and is characterised by a relapsing and remitting course.

Diagnosis of ulcerative colitis (UC) is made by the typical colonoscopic and pathological findings in patients who present with chronic diarrhoea. Colonoscopic findings in active disease include erythema, hyperemia, loss of vascularity, granularity, mucopurulent exudates, fragility, and ulcers in a continuous manner. However, these findings are not specific and repeated colonoscopy may be required.

If the inflammatory bowel diseases subclass is not available in the list, select Other autoimmune bowel disease and specify the disease name in the textbox in English.

Other Autoimmune/autoinflammatory diseases:

Select one of the options from the list:

Insulin-dependent diabetes mellitus (IDDM)

Type 1 diabetes mellitus is a metabolic disorder characterised by hyperglycemia due to absolute insulin deficiency. The condition develops due to the destruction of pancreatic beta cells, mostly by immune-mediated mechanisms. In some patients, there may be no evidence of autoimmune destruction of pancreatic beta cells; this is called idiopathic type 1 diabetes.

Diagnostic criteria for diabetes: fasting plasma glucose ≥ 7.0 mmol/L or 2-hour post-load plasma glucose ≥ 11.1 mmol/L or Hba1c ≥ 48 mmol/mol.[1]

If the disease subclass is not available in the list, select Other autoimmune disease and specify the disease name in the textbox in English.

VEXAS syndrome

VEXAS syndrome is an adult-onset autoinflammatory disease primarily affecting males, caused by a somatic mutation of the UBA1 gene in hematopoietic progenitor cells. The disease arises in late adulthood (typically after the age of 50) and causes both autoinflammatory and hematologic symptoms.

Which form of VEXAS syndrome

Please indicate the presence or absence of concurrent myelodysplastic neoplasms (MDS) at the time of VEXAS diagnosis or during the disease course.

[1] https://apps.who.int/iris/rest/bitstreams/1233344/retrieve

if VEXASsyndrome occurs with concurrent MDS, please complete the MDS diagnosis form and report VEXAS syndrome in Non-indication diagnosis form.

How was the VEXAS diagnosis made

Select the option that best describes how the diagnosis of VEXAS syndrome was made. If multiple diagnostic elements contributed, select the option that represents the most definitive method used.

Select **Clinical diagnosis only** if the diagnosis was made based on clinical presentation and supportive laboratory or imaging findings, without molecular confirmation of a UBA1 mutation and without definitive bone marrow diagnostic features.

Select **Confirmed UBA1 mutation (e.g. Met41)** if genetic testing confirmed a somatic mutation in the UBA1 gene, such as a mutation affecting Met41 or another pathogenic variant associated with VEXAS syndrome.

Select **Bone marrow features (e.g. vacuoles, dysplasia)** if the diagnosis was supported primarily by characteristic bone marrow findings, such as cytoplasmic vacuoles in myeloid and erythroid precursor cells, with or without dysplastic features, suggestive of VEXAS syndrome.

If the diagnosis was made using another diagnostic approach not listed above. Provide a brief description of the method used.

Previous Therapies

Previous therapy lines before HCT or CT

This section of the form is to specify which treatment the patient received since diagnosis. The treatments considered here use disease–modifying drugs, or drugs that are specifically given to delay the progression of the disease.

Indicate if the patient received any therapy before the current treatment by selecting **No** or **Yes**.

Systemic Sclerosis:

Among disease-modifying therapies currently used to treat SSc are immunosuppressants such as methotrexate, azathioprine, cyclophosphamide, mycophenolate mofetil, anti-fibrotic agents, such as nintedanib, and biologic therapies like rituximab (anti CD20 antibodies) or tocilizumab.

Systemic Lupus Erythematosus:

According to 2019 EULAR recommendations for SLE the following therapies are part of the treatment algorithm: corticosteroids, antimalarial drugs (mostly hydroxychloroquine), azathioprine, methotrexate, mycophenolate mofetil, cyclophosphamide, cyclosporine A and the biologic DMT with belimumab. Just recently, the bDMT anifrolumab and the calcineurin inhibitor voclosporine received approval from FDA and EMA.

Multiple sclerosis:

Over the last two decades, an increasing number of disease-modifying therapies (DMTs) have become available for the treatment of MS, therapies targeting various mechanisms including immune modulation, immune cell suppression or depletion and enhanced immune cell sequestration.

To date, various DMTs have been used with varying efficacy and safety profiles: interferon beta-1a (IFNb-1a), IFNb-1b, glatiramer acetate, mitoxantrone, alemtuzumab, teriflunomide, fingolimod, siponimod, ocrelizumab, natalizumab, ofatumumab, cladribine, dimethyl fumarate.

Emerging therapies include CNS-penetrant Bruton's tyrosine kinase inhibitors and therapies aimed at remyelination or neuroprotection.

Drugs

If you answered **Yes** to the previous question, also indicate for each agent in the list (chemotherapy, antibodies, etc.) if it was given or not to the patient as a part of the previous therapy by selecting **Yes**, **No** or **Unknown** next to each drug in the list.

If the drug/agent is not available on the list, select **Other** and report the generic drug/agent name(s) in the textbox in English.

Please consult the **LIST OF CHEMOTHERAPY DRUGS/AGENTS AND REGIMENS** on the EBMT website for drug/agent names. This document provides alternative names for many of the drugs/agents. Once you have found the drug/agent of interest on the list, add its database name to the table.

Dose

For cyclophosphamide, also report the total cumulative dose in mg.

Plasmapheresis

If you answered **Yes** to Previous therapy lines before HCT or CT, also indicate if the patient underwent plasmapheresis by selecting **Yes** or **No.** If this information is unavailable, select **Unknown**.

Previous surgical procedures

If you answered **Yes** to Previous therapy lines before HCT or CT and only for Crohn's disease, indicate if the patient underwent any previous surgical procedure by selecting **Yes** or **No**. If this information is unavailable, select **Unknown**.

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