Chapter 6
Principles of Assessment in Adolescent and Young Adult Rheumatology Practice

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Introduction

Musculoskeletal (MSK) presentations are common, ranking highly in the self-reported health problems amongst adolescents [7], and result from a spectrum of causes (Table 6.1), which is different to adults. In many cases the course is self-limiting, but it is important to consider potentially life-threatening 'red flag' conditions (e.g. malignancy, sepsis, vasculitis) or MSK associations with chronic conditions (e.g. inflammatory

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Table 6.1 Differential diagnosis of musculoskeletal pain

Differential diagnosis of joint pain
Life-threatening conditions
Malignancy (leukaemia, lymphoma, bone tumour)
Sepsis (septic arthritis, osteomyelitis)
Nonaccidental injury
Joint pain with no joint swelling
Hypermobility syndromes (patients sometimes report transient swelling)
Complex regional pain syndromes (localized or widespread)
Orthopaedic syndromes (e.g. slipped upper femoral epiphysis, Perthes' disease)
Metabolic (e.g. hypothyroidism, lysosomal storage diseases)
Referred pain from extra-articular causes (e.g. testicular torsion, inguinal hernia, urine infection, spinal tumour and nerve irritation)
Joint pain with joint swelling
JIA
Trauma (haemarthrosis)
Infection
Septic arthritis and osteomyelitis (viral, bacterial [including Lyme disease], mycobacterial)
Reactive arthritis (post-enteric, sexually acquired)
Infection-related (rheumatic fever, vaccination-related)
Inflammatory bowel disease (Crohn's disease, ulcerative colitis)
Autoimmune rheumatic disease (systemic lupus erythematosus, scleroderma, dermatomyositis)
– Joint swelling may be minimal (arthralgia > arthritis)
Sarcoidosis
Metabolic (e.g. osteomalacia [rickets], cystic fibrosis, mucopolysaccharidoses)
Haematological (haemophilia, haemoglobinopathy)
Tumour (benign/malignant)
Developmental/congenital (e.g. spondylo-epiphyseal dysplasia)
Chromosomal (e.g. Down's-related arthritis)

bowel disease, cystic fibrosis, psoriasis). The evidence-based approach to paediatric MSK clinical examination includes a basic examination (pGALS) [4] and a more detailed regional MSK examination (pREMS) [3]. The MSK assessment must be interpreted in the context of general assessment, suspected 'red flags' and systemic enquiry (Table 6.2). The important issues to highlight are described below and further illustrated with clinical cases.

The Impact of Growth

Normal variants (of age-related growth) in early childhood remain relevant to the AYA as persistent changes beyond accepted age ranges may present with functional problems (e.g. gait abnormality). Orthopaedic conditions (at the hip in particular) need to be excluded, e.g. persistent femoral anteversion presenting with in-toeing, tarsal coalition and the painful nonmobile flat foot. Joint hypermobility is common and may associate with non-specific MSK pain (see Chap. 11).

Localized growth abnormalities also occur and may indeed become more severe in adolescence, e.g. scoliosis from a primary defect in the spine or secondary to leg malalignment from leg length inequality and valgus deformity at the knee (from chronic untreated arthritis), micrognathia and shortened digits.

Pattern Recognition

The spectrum of MSK pathology varies with age, such as causes of limp (Table 6.3). The differential diagnosis of the acute limp must include orthopaedic conditions; slipped upper femoral epiphysis is more common in adolescents (especially if overweight). Chronic limp may result from missed developmental dysplasia of the hip, chronic untreated

Table 6.2 General examination in the context of musculoskeletal disease

	Features	Context and comments
General observations	Overall impression of Well-being Appearance, demeanour and interaction with parent or carer Features of dysmorphism (including faces and limbs/trunk/limb proportions) Height and weight plotted on a growth chart Pubertal status	An unwell AYA will require prompt admission and assessment for malignancy or sepsis Local safeguarding policies should be followed if concerned about nonaccidental injury or neglect Faltering growth may be a sign of systemic or chronic disease Short stature or localized growth problems are features of chronic untreated disease or dwarfism syndromes; abnormal trunk and limb proportions may not be apparent until final skeletal height is achieved (e.g. marfanoid syndromes)
Skin inspection	Look for rashes including scalp and hair Overall feel of the skin, including colour and texture	Skin psoriasis – Extensor surfaces, natal cleft Skin elasticity and scars suggestive of heritable collagen disorders If low mood, evidence of self-harming behaviour especially upper, non-dominant arms and thighs Malar butterfly rash – JSLE or JDMS Violaceous heliotrope rash or Gottron's papules on the hands – JDMS Evanescent macular salmon pink rash may be seen in systemic-onset JIA (often occurs with spikes of fever) and may demonstrate Koebner phenomenon Localized scleroderma may present with an isolated patch of pigmented skin (morphoea) – Systemic sclerosis is rare in childhood Vasculitis or livedo rash may occur in connective tissue disease (including JSLE or JDMS)

Nail examination	Nail pitting (psoriasis) Nail beds and capillaroscopy can be aided by magnification using a gel and ophthalmoscope or dermascope	Nail change of psoriasis may be subtle and the only manifestation of psoriasis Dilated, tortuous nail bed capillaries suggest active inflammation in the context of connective tissue disease
Ear, nose and throat examination	Cervical lymphadenopathy Oral mucosa, gums and teeth Ears and nose (bridge and mucosa) Parotid swelling if sicca features or suspicion of connective tissue disease or sarcoidosis	Significant cervical lymphadenopathy may occur in malignancy or multisystem disease Mouth ulcers – JSLE, inflammatory bowel disease and Bechet's disease. Perianal or genital examination may be required Sjögren's syndrome may be a feature within mixed connective tissue disease Poor dental hygiene is a concern, particularly in the immunosuppressed AYA, and may be more common in JIA with temporomandibular involvement or micrognathia ENT abnormalities are common in ANCA +ve vasculitis, e.g. saddle nose, sinusitis (granulomatosis with polyangiitis)
Cardiovascular	Blood pressure and pulses Presence of bruits Heart sounds	Hypertension in the context of rheumatic disease may signify renal involvement (e.g. vasculitic disease) Pericarditis is a feature of systemic-onset JIA and vasculitic disease Cardiac abnormalities are a feature of non-benign hypermobility syndromes (e.g. Marfan's and Ehlers–Danlos)
Respiratory	Lung fields Pulmonary function testing	Restrictive lung disease may be seen in connective tissue disease

(continued)

TABLE 6.2 (continued)

	Features	Context and comments
Abdominal	Presence of tenderness/guarding Bruits Hepatosplenomegaly	Multisystem disease, including vasculitis, malignancy or inflammatory bowel disease Inflammatory bowel disease may be indolent or suspected with NSAID intolerance
Neurological	Full neurological examination of the lower limbs is always indicated with back pain Cranial nerve assessment in the context of headache Peripheral nerve involvement muscle power	Abnormal neurological examination (such as altered sensation or hyper-reflexia) in back pain should lead to urgent imaging and expert assessment MRI ± angiography may be needed if cerebral vasculitis is suspected Reduced muscle strength is seen in JDMS and mixed connective tissue disease
Eye	Acute red eye Reduced visual acuity Fundoscopy Slit lamp examination	In the AYA with JIA, chronic anterior uveitis may be asymptomatic and only detected by eye screening. Acute anterior uveitis may present with photophobia and a painful red eye as a feature of HLA-B27-related JIA/ERA Multisystem disease (e.g. sarcoidosis, vasculitis) may manifest with ocular involvement
Renal	Blood pressure Urinalysis	Haematuria is a feature of renal disease associated with ANCA +ve vasculitis (i.e. Wegener's granulomatosis) JSLE-associated nephritis may present with hypertension and proteinuria Exclusion of UTI is important particularly if immunosuppressed

ANCA anti-neutrophil cytoplasmic antibodies, *ENT* ear, nose and throat, *JDMS* juvenile dermatomyositis, *JIA* juvenile idiopathic arthritis, *JSLE* juvenile systemic lupus erythematosus, *UTI* urinary tract infection

Table 6.3 Significant causes of limp

	4–10 years	11–16 years
Most common	Trauma Transient synovitis Perthes' disease	Trauma Osgood–Schlatter disease
Conditions requiring urgent intervention	Osteomyelitis Septic arthritis	Osteomyelitis Septic arthritis
	Nonaccidental injury Malignant disease (e.g. acute lymphocytic leukaemia)	Slipped upper femoral epiphysis Malignancy (e.g. bone tumours)
	Testicular torsion Appendicitis Inguinal hernia	
Other important conditions to consider at all ages	JIA Late presentation of developmental dysplasia of the hip Metabolic (e.g. rickets) Haematological disease (e.g. sickle cell anaemia) Reactive arthritis Lyme arthritis	

JIA or previous joint disease (e.g. Perthes). Patterns of joint involvement and extra-articular features in JIA for the AYA differ from both adult rheumatoid arthritis and the archetypal child with JIA (see Chap. 7).

The History and the Physical Examination

The systematic enquiry into family and past history may be helpful (see Tables 6.1 and 6.2). A recent travel history (e.g. to an endemic area for Lyme disease) may be informative,

and a sexual history is important in the adolescent with reactive arthritis and needs to be explored sensitively, acknowledging safeguarding concerns and the need for privacy and confidentiality. A medication history (e.g. response to NSAIDs, potential for drug-induced lupus), family history (e.g. autoimmune disease, muscle disease), social history and family dynamics may be also revealing. It is important to probe for symptoms suggestive of inflammatory joint or muscle disease (e.g. morning stiffness, gelling after rest, swelling) and discriminate against mechanical causes of pain (e.g. locking or giving way, pain worse on weight bearing or exercise). Conversely non-specific aches and pains are a feature of idiopathic pain syndromes (see Chap. 12).

Red flags (such as fever, malaise, anorexia, weight loss, bone pain, persistent night waking or raised inflammatory markers) are suggestive of serious potentially life-threatening conditions (e.g. sepsis or malignancy or multisystem disease), and a full general examination is needed. Additional clinical skills pending the clinical scenario include nailfold capillaroscopy or muscle strength testing (if connective tissue or muscle disease suspected, respectively).

MSK history-taking can be misleading, even when taken by experienced clinicians, as the history alone may not identify sites of joint involvement [5]. It is important, therefore, that in all cases where MSK disease is suspected, a basic joint examination (such as pGALS; see later) is needed, as a minimum, to assess all joints. In the examination of AYA with learning disabilities and complex medical needs, joint problems can easily be overlooked; there is an association of inflammatory joint disease with chromosomal disorders (such as Down's syndrome or DiGeorge syndrome), and many complex genetic conditions (such as mucopolysaccharidoses) may present with or develop joint problems that can be indolent in older children/AYA [2, 11].

pGALS and pREMS Musculoskeletal Examinations

The paediatric gait, arms, legs, spine (pGALS) assessment [4] is a simple, quick, evidence-based validated approach to MSK assessment in children based on the adult GALS (gait, arms, legs, spine) screen, with a series of simple manoeuvres. pGALS is useful to identify abnormal joints (e.g. inflammatory arthritis, orthopaedic problems at the hip, scoliosis). Following pGALS, the observer is directed to a more detailed examination of the relevant area(s) using the consensus approach to paediatric regional examination of the musculoskeletal system (called pREMS) [3]. This is based on the 'look, feel, move, function' principle similar to that of adult REMS for each joint or anatomical region, although there are differences reflecting pathologies from those observed in adults and the influence of growth and development; some tests (e.g. Schober's test for lumbosacral movement) are not reliable in younger adolescents; a history of inflammatory back symptoms in the presence of equivocal physical findings may necessitate imaging (MRI) of the spine and sacroiliac joints (Chap. 14).

There is no absolute age 'cut-off' as to when to use pGALS or GALS or indeed pREMS or REMS. However, we would recommend that pGALS and pREMS be used in the younger adolescents and for those with chronic disease that started in childhood, given the differences in patterns of joint involvement in JIA and spectrum of pathology in AYA compared to adults.

Along with general considerations in clinical consultations with AYA such as communication skills and respecting confidentiality (see Chap. 4), MSK clinical assessment also requires:

- An appropriate environment for AYA to be assessed. The appointment letter can explain that examination is important and suggest that appropriate clothing is brought to the appointment.
- Consideration of a chaperone (other than the accompanying parent/carer/friend/partner).

Full details of the examination techniques with video demonstrations are provided on the PMM website [12] and in Further Reading and Resources.

Training in Clinical Assessment of the AYA

Traditionally adult and paediatric clinical skills are taught independently, but it is important that all healthcare professionals have an appreciation of the additional complexity of the clinical assessment of AYA, as this group will present to primary care and either adult or paediatric services (see Chaps. 1, 2, 3, and 4). Acquisition of 'core' MSK clinical skills for paediatrics and adult practice [1, 6, 8–10] needs to start within undergraduate education as the foundation for further postgraduate training.

Clinical Scenario to Illustrate Key Differences from Paediatric and Adult Clinical Assessment

Javeen is a 15-year-old boy, born in the UK with parents from India. He attends with his adult sister as his parents are at work and presents with a sore left ankle and foot for 2–3 weeks. He initially ascribed his symptoms to soccer playing although he cannot recall a specific injury. In the last week, his pain is present in the mornings and also later as the day progresses. He is well in himself, more tired recently. There is no specific family history or past history of note. He has no history of red eyes.

On MSK examination he has difficulty on heel walking on the left side, and his left calf is slightly wasted. His lumbar spine appears restricted on forward flexion (he cannot reach his knees), and he has a nonmobile flat foot on the left, restricted midfoot movement and tenderness around the left heel. His spine has no tenderness, and he has a good range of movement with a normal Schober's measurement (15 cm

increasing to 21 cm). Joint examination is otherwise normal, and he has no enthesitis and tender points elsewhere, nor skin abnormality.

Diagnosis Javeen has enthesitis-related arthritis. He is HLA-B27 positive.

Learning Points

- With a pattern of oligo-JIA in adolescence, it is important to consider ERA.
  - Enquire about family history (spondyloarthropathy, inflammatory bowel disease) and acute uveitis and check for enthesitis.
  - The history of pain on heel walking suggests enthesopathy.
  - Consider inflammatory joint disease with a nonmobile painful flat foot.
- Muscle wasting suggests prolonged disease duration and a more chronic aetiology.
- Schober's test is not reliable as an indicator of sacroiliac joint disease in AYA, and further imaging (MRI) may be needed.

Key Management Points

1. Musculoskeletal presentations are common during adolescence and young adulthood.
2. The impact of growth, localised and generalised, should be considered during clinical assessment of adolescents
3. Pattern recognition for the AYA differs from both adult and archetypal paediatric diseases and should be considered in the context of the young person's developmental stage

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Further Reading and Resources

BSR eLearning modules – https://rheumatologylearning.com – modules on adult JIA targeting educational needs of adult Rheumatology healthcare professionals.

E-Learning for Healthcare. https://www.e-lfh.org.uk – modules on limp, normal Development and adolescent health issues.

EULAR/PReS Online course in Paediatric rheumatology https://www.eular.org/edu_online_course_paediatric.cfm.

EUTEACH – https://www.unil.ch/euteach/en/home.html – European Training in Effective Adolescent care and health.

Newcastle University short online courses – paediatric musculoskeletal modules – www.cpd.ncl.ac.uk.

Paediatric musculoskeletal matters[12] – www.pmmonline.org.

pGALS app – a free resource. Available through app stores and in multiple languages.