Spinal Muscular Atrophy:
Adult Care Pathway

ENTER

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About this pathway

This integrated care pathway has been created as a collaborative effort – our special thanks to the spinal muscular atrophy (SMA) professionals representing a range of specialist SMA centres who contributed to this project. We hope the pathway will be a useful resource locally for both healthcare professionals and providers to map the SMA journey and best practice care. We are particularly keen to share this resource and receive comments in order to ensure the pathway is as comprehensive as possible.

Currently awareness of SMA and access to SMA services across the UK is variable. As a result many individuals may wait too long for reviews and assessment which can have major ramifications for their symptom management and quality of life. It is essential that the complexity of SMA care is understood in order to make the most meaningful improvements to local services. We hope this integrated care pathway will help to unravel the SMA journey and aid you in improving and streamlining the care delivered in your area.

Standards of care are already available but granularity around the patient journey is important for specialists and generalists so that they can understand the care adults with SMA are likely to need throughout their journey. If the pathway is explicit, then:

- Patient journey is smoother
- Teams can ensure the right care is delivered at the right time.

International Standards of Care

- Part 1: Diagnosis and management of spinal muscular atrophy: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care – ScienceDirect
- Part 2: Diagnosis and management of spinal muscular atrophy: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics - ScienceDirect

The SMA integrated care pathway is easy to navigate by clicking on the menu tabs.

Click on icons in the pathway to open further information:

Information

Red flag & alerts

Sub pathway

The pathway is designed to be viewed electronically. Some links redirect to resources that will open in your internet browser – these will require an internet connection.

About SMA

Spinal muscular atrophies (SMA) include a group of neuromuscular disorders characterised by degeneration of alpha motor neurons in the spinal cord with progressive muscle atrophy, weakness and paralysis. The most common form of SMA is due to a defect in the survival motor neuron 1 (SMN1) gene localised to 5q11.2-q13.3. It includes a wide range of phenotypes that are classified into clinical groups on the basis of age of onset and maximum motor function achieved.

Approximately 1 in 40 people carry an SMA-associated genetic mutation in the survival motor neurone 1 gene (SMN1) leading to an insufficient production of full length SMA protein. As a result 1:10,000 infants is born with the condition. The majority of infants with SMA have the most severe form of the disease (SMA I) in which symptoms manifest in the first few weeks or months of life. Milder forms of the disease also exist, e.g. SMA II, III and IV, which are associated with later onset and better prognosis. With available treatments the SMA phenotype classification is evolving towards a functional status definition:

Management of SMA

SMA is managed through multidisciplinary supportive care. Treatment should follow guidelines from the International Standards of Care Committee for Spinal Muscular Atrophy. Supportive care strategies aim to minimise the impact of disability, address complications and improve quality of life. These may involve respiratory, gastroenterology, and orthopaedic care, as well as nutritional support, physiotherapy, assistive technologies, occupational therapy and social care. New treatments now present opportunities to change the management and survival landscape of SMA.

Treatment expectations

Treatment expectations are subjective and based on each individual patient. To make sure the right treatment expectations are set a broad and holistic understanding of each patient's needs is required. These relate to: treatment access, access to supportive care, education level, geography, social isolation, information, peer-to-peer exchange and access to technology.

What does it mean for patients and carers?

- Functional improvement from baseline – any improvement is good. SMA is a deteriorating disease, so even stabilisation is seen as beneficial.
- Reduced manifestations.
- Improved life expectancy and stabilisation.

Treatment expectations for emerging phenotypes:

SMA I	SMA II	SMA III (childhood onset)	SMA IV (adulthood onset)

Shortened life expectancy without treatment — Normal life expectancy

Acknowledgements

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- Rebecca Flesher, Occupational therapist, The Walton Centre NHS Foundation Trust, Liverpool
- Dr Clare Galtrey, Consultant neurologist, St George's Hospital London
- Dr Channa Hewamadduma, Neuromuscular physician, Sheffield Teaching Hospitals NHS Foundation Trust
- Natasha Hoyle, Neurosciences pharmacist, Sheffield Teaching Hospitals NHS Foundation Trust
- Dr James Lilleker, Neurology consultant, Salford Royal NHS Foundation Trust, Salford
- Lindsay Maidment, Neuromuscular clinical specialist physiotherapist, Sheffield Teaching Hospitals NHS Foundation Trust
- Dr Chiara Marini-Bettolo, Consultant neurologist, John Walton Muscular Dystrophy Research Centre, Newcastle
- Dr Andria Merrison, Consultant neurologist, North Bristol NHS Trust, Bristol
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- Sun Narayan, Neuromuscular specialist physiotherapist and clinical coordinator, University Hospital Southampton NHS Foundation Trust

Facilitator

- Sue Thomas, Independent Healthcare Consultant

Pathway design

- Sarah Mehta, Medical Writer

Centres involved in the development of this pathway:

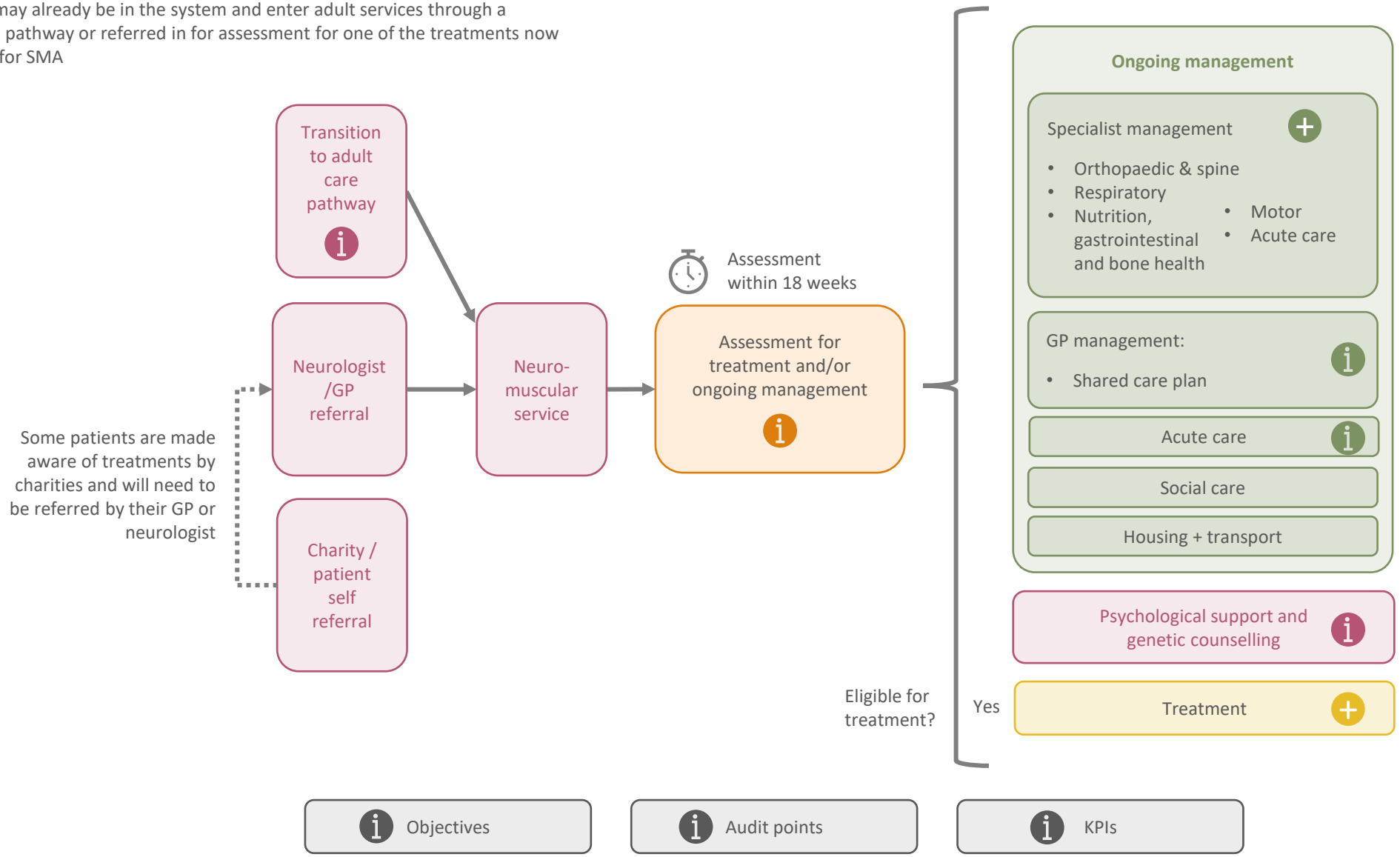
- Cardiff and Value University Health Board
- John Walton MND Research Centre, Newcastle
- Sheffield Teaching Hospitals
- Salford Royal NHS Foundation Trust
- St Georges Hospital, London
- The Walton Centre NHS Foundation Trust, Liverpool
- University Hospital Southampton NHS Foundation Trust

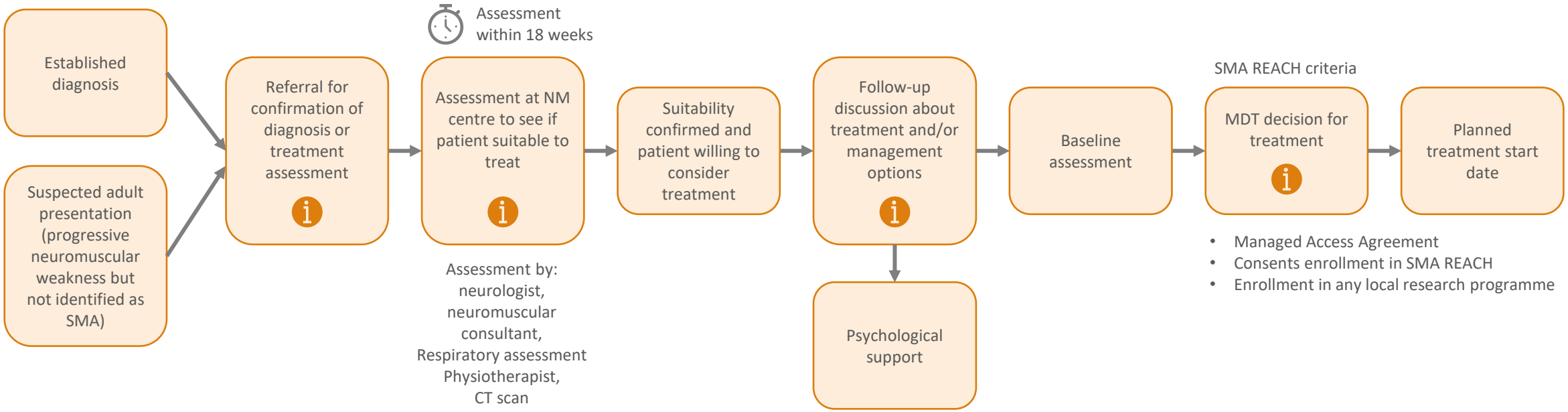
Contact us

Please contact us with your comments and feedback at: info@neurologyacademy.org

Document due for review in November 2021.

Patients may already be in the system and enter adult services through a transition pathway or referred in for assessment for one of the treatments now available for SMA





- Adult onset ambulatory (Type IV). Symptom onset aged 19 or older. The patient takes at least five steps independently in the upright position with the back straight. One leg moves forward while the other supports most of the body weight. There is no contact with a person or object.
- “Walking unaided” (“ambulant” i.e. type III SMA patient): Symptom onset aged 18 or younger. The patient takes at least five steps independently in the upright position with the back straight. One leg moves forward while the other supports most of the body weight. There is no contact with a person or object.
- “Sitting without support” (“sitter” i.e. type II SMA): The patient sits up straight with the head erect for at least 10 seconds. The patient does not use arms or hands to balance body or support position.

Nusinersen (Spinraza)

Nusinersen is provided under a Managed Access Agreement (MAA). The drug is an antisense oligonucleotide drug that modifies pre-messenger RNA splicing of the SMN2 gene and thus promotes increased production of full-length, more functional SMN protein. Administered intrathecally, it is the first drug to have been licensed for the treatment of 5q SMA. Clinical trials have shown significant improvement in motor function with children who have been treated enabling them to achieve motor milestones that are unprecedented in the natural history of the condition. The drug has demonstrated efficacy in treated children and clinical trials have also highlighted that there may be a benefit in patients who start treatment earlier.

In July 2019, NICE, Biogen and NHS England came to an arrangement for the MAA, which allows children and adults with SMA Types I, II and III to have the treatment in England if they meet access criteria. The MAA is an interim scheme that enables data collection on treatment effectiveness whilst ensuring treatment access. The Spinraza agreement has been granted for five years and currently runs to 2024.

- NICE (2019) MAA for nusinersen
- NICE (2019) Nusinersen for treating spinal muscular atrophy. Technology appraisal guidance [TA588]

Risdiplam (Evrysdi)

Risdiplam is provided under the Early Access to Medicines Scheme (EAMS). This drug is a survival motor neuron 2 (SMN2) splicing modifier designed to treat SMA caused by mutations in chromosome 5q that lead to SMN protein deficiency. This small molecule targets and encourages the SMN2 ‘back-up’ gene to produce a greater amount of functional SMN protein, which is lacking in people with SMA.

Risdiplam is being studied in a broad range of patients who have SMA from birth to 60 years of age. It is an orally-administered liquid designed to provide a sustained increase in SMN protein centrally and peripherally when given daily at home in liquid form by mouth or by feeding tube making it suitable for when patients may not be able to tolerate intrathecal injections or eligible for nusinersen.

- NICE technology appraisal: Risdiplam for treating spinal muscular atrophy in children and adults (publication expected July 2021)
- Further information about EAMS

- The EAMS scheme is now closed to new patients following marketing authorisation from the MHRA (May 2021). Existing patients participating in the EAMS can continue to be treated until reimbursement.

Referral to SMA clinic – request old notes and genetic report

Video consultation with consultant, physio and nurse

Face-to-face consultation*:
- 30 mins consultant: examination and consent.
- 2 hour assessment with physio and nurse: bloods**, urine protein, sniff nasal-inspiratory force, XR lumbar spine or CT spine.
- SMA Reach data input 30 mins (research nurse or physio).
- Referral to respiratory ventilation clinic if no up-to-date assessment.

*Ensure beforehand all notes and results available and all necessary investigations booked on day and explanation to patient of expectations of visit.
**FBC, U and E, renal function and clotting +/- genetic test to confirm SMN1 mutation and SMN2 copy number.

Patient & MDT* decision for treatment (or continue with existing treatment regime)

*Neuromuscular consultant x 2, neuroradiology consultant & neuromuscular physiotherapist

Eligible for nusinersen treatment?

No → Risdiplam oral medication

Yes

- Plan treatment start date
- Liaise with NDU & nusinersen nurse
- Consent
- Scans for administration
- Liaise with pharmacy
- Order drug
- Blueteq forms

Register with SMA Reach database

Baseline assessment: spinal imaging, bloods

NDU day case admission: Loading dose by intrathecal infusion

Comply with reporting data, assess patient

Second dose Day 14 (day case)

Third dose Day 28 (day case)

Fourth dose Day 63 (day case)

Main-tenance dose at 6 months (day case)

Clinical assessment: 30 mins consultant for examination and consent, then 2 hour assessment with physio and nurse, sniff nasal-inspiratory force measurement

Assess-ment every 6 months (out-patient)

Main-tenance dose at 10 months (day case)

Main-tenance dose at 14 months (day case)

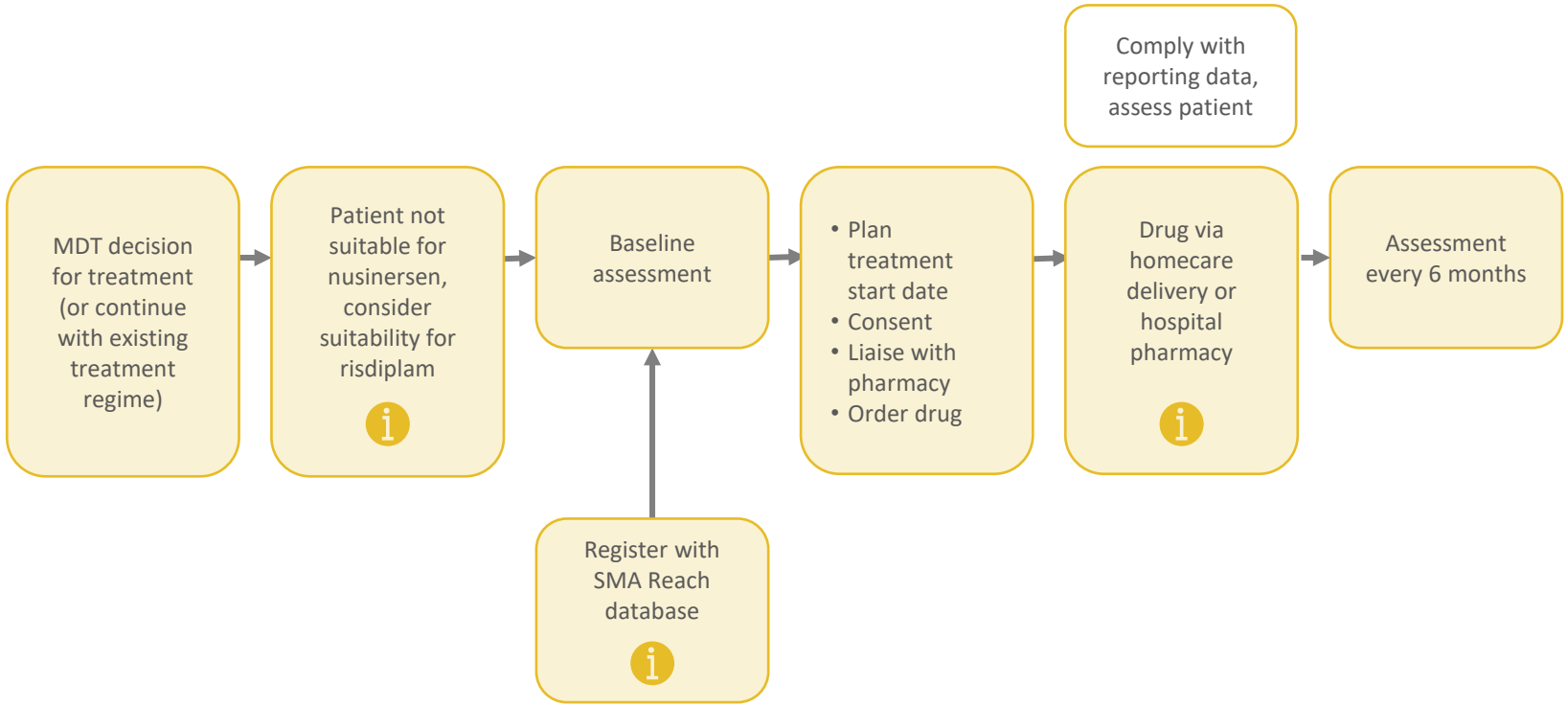
MDT review at 12 months to assess stabilisation, decline or improvement

*Interventional neuro-radiologist fluoroscopic LP imaging in some cases and 4 hour observation in NDU (patients that need hoist transfer to be admitted night before for overnight stay)

Follow-up phone nurse appointment after 1 week to check patient re outcomes & side effects

Arrange FBC and clotting renal function and urine protein for next dose

(If assessment cannot be done when the patient attends for infusion this may result in an extra visit)
NDU = neurology day unit


MDT decision for treatment (or continue with existing treatment regime)
Patient not suitable for nusinersen, consider suitability for risdiplam
Baseline assessment
Register with SMA Reach database
• Plan treatment start date
• Consent
• Liaise with pharmacy
• Order drug
Comply with reporting data, assess patient
Drug via homecare delivery or hospital pharmacy
Assessment every 6 months

Neuromuscular management

A multidisciplinary team approach is the key element in management of people with SMA and the different aspects of care should not be dealt with isolation but as part of a MDT approach. A coordinator should be available to coordinate all aspects of care. The diagnosis and management of SMA recommendations for care highlight an approach for management that addresses the needs of:

- Non-sitters
- Sitters
- Ambulant patients.

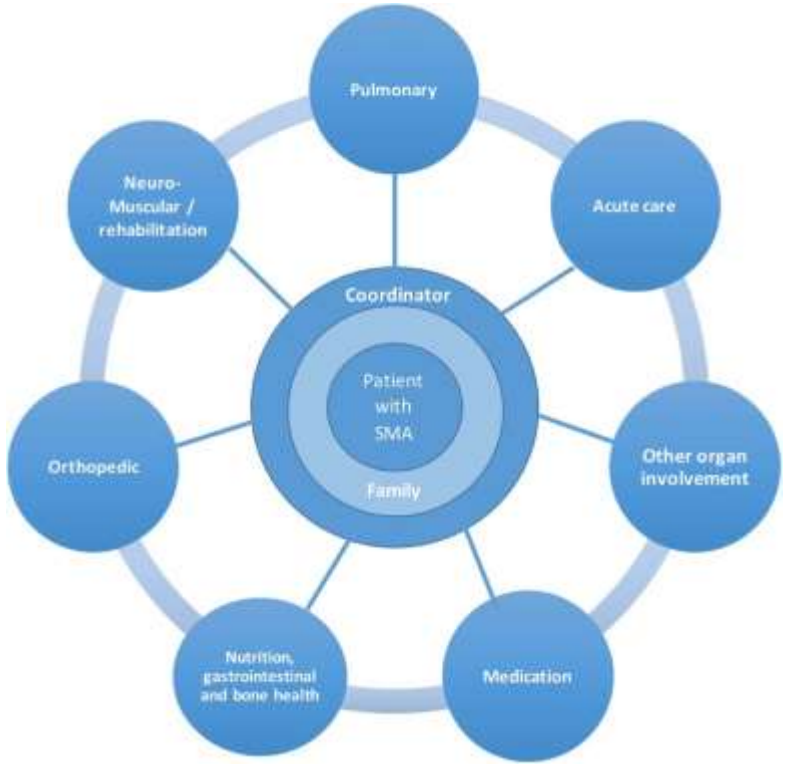
The key to all management is a baseline assessment and regular review of the person's individual ability (see SMA outcome measures and measures evaluated in clinical trials). Any improvement is good.

Rehabilitation: physiotherapy, occupational therapy, speech and language therapy

- Contracture management
- Movement abilitation
- Equipment/adaptations
- Swallow management.

Respiratory rehabilitation and management:

- Baseline respiratory assessments
- Individualised respiratory care plans
- Routine follow up including sleep studies
- When appropriate, use BiPAP and/or cough assist
- Training on management of emergencies.



Multidisciplinary approach.
Source: Mercuri E, Finkel RS, Muntoni F et al (2017) Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care

Orthopaedic management & impact on spine

Regular assessment and review: orthopaedic consultant, physio and OT

Non-sitters

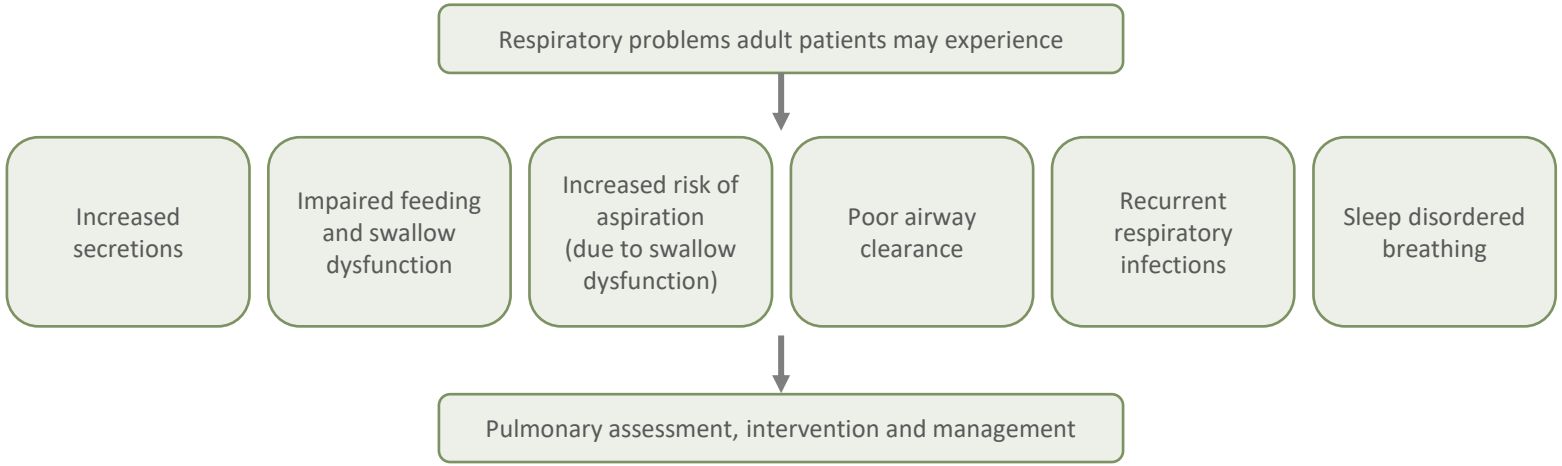
- Reduce impact on tight joints
- Prevent scoliosis
- Postural control and bracing
- Customised upright seating, standing frames
- Thoracic and cervical bracing
- Modifications should include respiratory support.
  Ensure no restrictions on thoracic expansion / secretion clearance
- Stretching
- Muscle weakness: promote functioning
- Communication
- Cobb angle >20° rechecked 6-monthly until bones no longer growing

Sitters

- Mobility aids – carry out assessment powered wheelchair / customised seating / vehicle

Ambulant

- Promote function & mobility
- Exercise, e.g. swimming
- Contractures: stretching
- Posture: position & bracing
- Promote function & mobility, lightweight wheelchair, accessible driving


Respiratory problems adult patients may experience
Increased secretions
Impaired feeding and swallow dysfunction
Increased risk of aspiration (due to swallow dysfunction)
Poor airway clearance
Recurrent respiratory infections
Sleep disordered breathing
Pulmonary assessment, intervention and management

Next »

Pulmonary assessment, intervention and management:

Non-sitters	Sitters	Ambulant
• Assessment of hypoventilation (end tidal CO_2). • Sleep study or pneumograms in all symptomatic patients or to determine if NIV initiation needed. • Assessment of gastroesophageal reflux.	• Spirometry • Sleep study or pneumograms when minimal suspicion of symptoms of nocturnal hypoventilation. • Assessment of gastroesophageal reflux.	• Review of cough effectiveness. • Detailed search for signs of nocturnal hypoventilation.
Support airway clearance: • Oral suctioning. • Physiotherapy/respiratory therapy should be implemented immediately: Manual chest therapy. • Cough insufflator/exsufflator.	Nebulised bronchodilators with asthma or a positive bronchodilator response RSV vaccine, influenza vaccination annually	Nebulised bronchodilators in patients with suspicion of asthma immunisations, annual influenza and pneumococcal vaccination.
• Assessments 3 months initially, then 6 monthly. • Airway clearance with physiotherapy/respiratory therapy. • Cough assist essential with ineffective cough.	Immediate physiotherapy/respiratory therapy: Manual chest physiotherapy Cough insufflator/exsufflator Assessments 6 monthly, airway clearance is critical with ineffective cough.	Mucolytics should not be used long-term. Referral to respiratory team if: • Evidence of weak cough • Recurrent infections • Suspicion of nocturnal hypoventilation
Support ventilation with bilevel NIV in symptomatic patients.		
Ventilation should be started in all symptomatic patients. Some experts recommend using it during acute respiratory illnesses to facilitate discharge. NIV should be initiated during a sleep study or observing the patient clinically for adequate gas exchange. NIV interfaces should be fitted by skilled physiotherapists selecting two interfaces to alternate skin contact points.		

Source: Mercuri E, Finkel RS, Muntoni F et al (2017) Diagnosis and management of spinal muscular atrophy

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Physiotherapy and rehabilitation

Non sitters

Physiotherapy and rehabilitation aims to reduce impact on tight joints, optimise function and help a patient tolerate different positions, lying or sitting with assistance. Regular assessment from physiotherapist and occupational therapist (OT) is required.

- Positioning, bean bags, wedges and pillows help support non-sitters.
- Custom made seats, reclining or sitting strollers and power chairs help provide support.
- **Stretching**: flexibility is important, utilise:
  - Assisted stretches.
  - Splints to support or immobilise limbs or spine
  - Splints should be applied for more than 60 minutes or overnight.
  - Braces used to support part of the body for stabilisation – these should be used at least 5 times a week.
- **Neck collars**: helpful for head support and to assist breathing.
- **Standing frames**: used to help maintain and improve posture. They also help with bone health and digestion.
- Exercise and movement in water can be helpful providing the head is well supported and the patient supervised.

Sitters

The main objective is to reduce impact and flexibility of tight joints and prevent scoliosis. physiotherapy and occupational therapy should give guidance and training on how to achieve aims.

- **Orthoses**: to support arms, leg and spine to assist movement or achieve activities such as standing and supported walking.
- **Braces**: to stabilise use minimum x 5 weekly.
- **Splints and braces**: keep joints in certain positions should be worn for 60 mins or overnight.
- **Neck support and supported standing**: stretches legs, promotes good posture increases bone density, blood circulation and eases constipation. 60 mins minimum 3–5 times weekly, 5–7 times is recommended
- **Stretching**: combining effective stretches with splints and standing exercises is crucial. Routine should be adapted individually by physiotherapy or occupational therapy 5–7 times a week.
- **Mobility and exercise**: all sitters should have a powered wheelchair and custom seating, mobility aids and an accessible car. Exercise will maintain and improve strength, flexibility, resilience and balance and improve participation in school and for leisure and social activities. Resistance training, swimming, horse riding and wheelchair sports are all useful ways to participate in exercise.

Aambulant

Involvement of physiotherapy and occupational therapy.

The overall aim is to promote maximum functional independence in day-to-day activities including work, education and recreation. Maintain strength, mobility, balance and range of movement:

- Work on flexibility strength endurance and balance.
- Stretching.
- Positioning.
- Mobility and balance exercise.
- Lightweight manual wheelchairs or ones with power-assisted wheels useful as may be tiring to walk.
- Powered wheelchairs or scooters for long distance.
- Accessible vehicles.
- Ensure accessible environment at home, education and work.
- Improve participation.
- Encourage leisure activities.

Key messages

- Safe swallowing is one of the most important aspects of care as patients with a weak swallow are at risk of inhaling (aspirating) their food which can cause choking and chest (respiratory) infections.
- Review with speech and language therapist annually.
- Standards of Care recommend that a dietician reviews feeding and diet annually.
- If swallowing becomes unsafe, or if the patient is not maintaining their weight, feeding alternatives may be suggested:
    - Short-term options may include feeding through a:
        - Nasogastric (NG) tube - a thin flexible feeding tube passed through the nose into the stomach.
        - Nasojejunal (NJ) tube - through the nose into the middle part of the small intestine (the jejunum).
    - A longer-term option is:
        - Gastrostomy (PEG) tube - placed in the stomach via a surgical procedure and also called a PEG - percutaneous endoscopic gastrostomy. Another procedure called a Nissen Fundoplication, which helps to reduce any reflux, may be done at the same time.

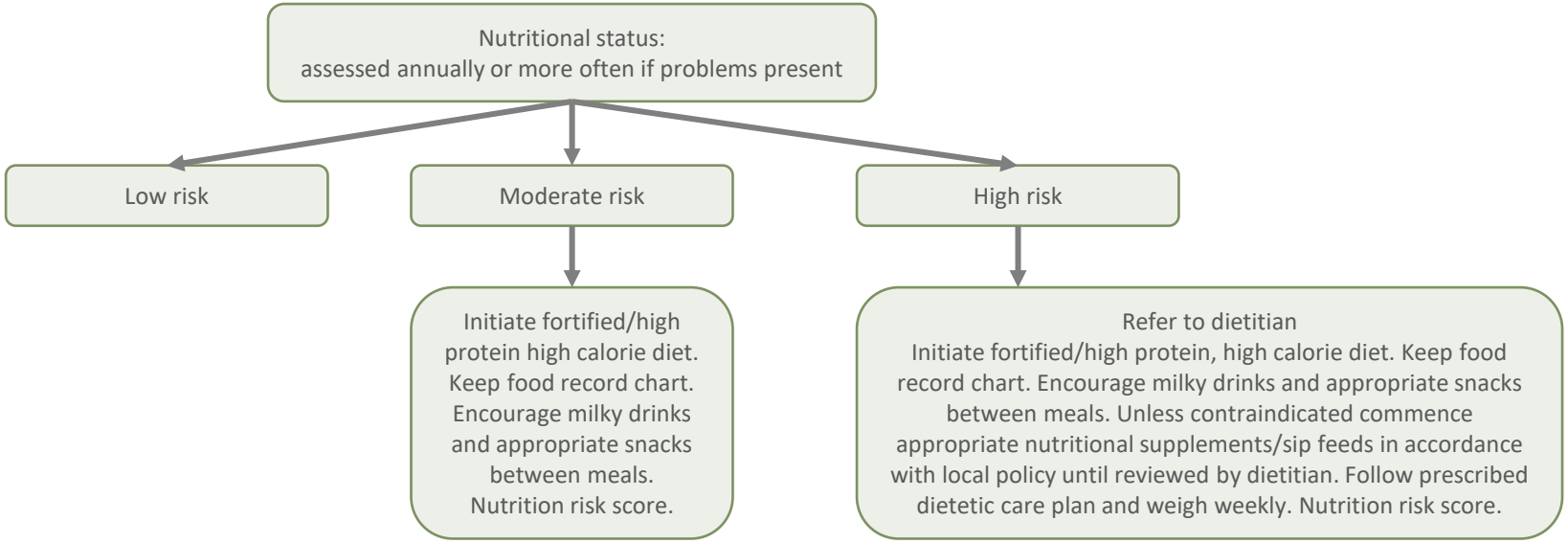
Next »

Nutritional assessment, swallowing and gastrointestinal dysfunction and intervention

Non-sitters	Sitters	Aambulant
Assessment safe swallowing – bulbar dysfunction can result in aspiration and pulmonary infections.	Assessment of symptoms of dysphagia/aspiration/difficulties with feeding. Video fluoroscopic swallow study if clinical signs suggestive of dysphagia. Nutritional analysis of food records/feeding regimen, longitudinal anthropometrics (height, weight, OFC). Nutrition tabs may be indicated. Acute care monitoring glucose metabolism labs 25 hydroxy-vitamin D tabs, body composition and bone density (DXA).	Dietician assessment • See dietitian if over/under nutrition • Nutritional analysis/monitoring if underweight or overweight • Longitudinal anthropometrics (height, weight, BMI, OFC). Glucose metabolism labs 25 hydroxy-vitamin D tablets.
Oral feeding may be limited.	Constipation management – evaluation of fluid and fibre intake recommended for constipation	Bone health important • High incidence of osteopenia and fractures • DEXA scan
Interventions – failed swallow study NG tube or NJ tube.	Dietician evaluation for concerns of under/over nutrition Possibility of obesity check BMI. Dietician assessment annually or more often if required. NB Evaluation important for specialised diets.	Speech and language assessment • Assessment of symptoms of dysphagia, aspiration, difficulties with feeding. • Video fluoroscopic swallow study if suggested by clinical signs suggestive of dysphagia. • Nutritional analysis of food records/feeding regimen. • Longitudinal anthropometrics (height, weight, OFC). • If failed swallow or interventions are not sufficient placement of a long-term gastric feeding tube. • If overweight, limit calorie intake and maximise nutrient intake. • Minimise fasting during acute care. Appropriate fasting time depends on prior nutritional status and nature of acute event. • Provide adequate fluid intake during illness. Monitor electrolyte levels and correct as needed.
Adequate hydration and electrolyte balance important during illness.	NB. People with SMA are at risk of hypoglycaemia; following period of vomiting or fasting ensure sufficient fluids to prevent ketosis.	
Video fluoroscopic swallow study shortly after diagnosis and when suggested by clinical signs suggestive of dysphagia (fatigue, humid voice, pneumonias). Difficulties with eating (pocketing, jaw contractures, increased mealtimes). Nutritional analysis of food records, longitudinal anthropometrics, acute care monitoring 25 hydroxy-vitamin D tabs, body composition and bone density.		
Care considerations: Determine appropriate calorie needs based on growth. Standardised growth charts to track growth trends in combination with body composition measurement tools to assess appropriate growth.		
Dietician evaluation annually. NB Evaluation essential important for those on specialised diets.		

Source: Mercuri E, Finkel RS, Muntoni F et al (2017) Diagnosis and management of spinal muscular atrophy

Nutritional care pathway



Monitor weight, height and BMI

Anyone with elevated BMI should be assessed for possible obesity / excess body fat. Body composition monitored to ensure proportion of bone fat and muscle is healthy. Reduced mobility may result in weight gain.

Swallowing

Rare for ambulant patients to have swallowing and feeding difficulties but non-sitters and sitters may have NG/NJ/gastric tube in situ. Ensure adequate nutrition.

Constipation

Can be an issue due to lack of mobility and diet

Hypoglycaemia

People with SMA are at risk of hypoglycaemia (low blood sugar) following periods of vomiting or fasting ensure sufficient fluids to prevent ketosis.

Signs of hypoglycaemia: pale, clammy, tired, confused, glazed, not acting as they normally should, unable to wake them.

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Acute care

Chest infections and breathing issues are the most frequent problems that require acute or emergency care.

There should be an emergency healthcare plan or illness plan in place written by the medical team including the following information:

- Brief summary of the individual's diagnosis/es and their understanding of it.
- What are the warning signs or indications that the patient should be taken to hospital?
- Which healthcare providers should be contacted in an emergency?
- A list of regular and PRN medications, and indications for any rescue medications left in the patient's home for emergency use.
- Any ceilings of care that have been requested by the patient and any that have been recommended by healthcare professionals.
- Describe actions for emergencies arising at home.
- Preferences around respiratory management and preferences for supported breathing i.e. NIV / intubation.
- Any neck or jaw limitations.
- Nutrition and fluids requirements.
- Techniques used for clearing secretions.
- When and which antibiotics should be given.
- Action agreed if resuscitation is required.
- Individual wishes of the patient.

Where possible, local emergency medical services should be contacted in advance to discussed any specific needs and what equipment is used at home. In an emergency, you should go to the closest hospital. Wherever possible, the equipment used at home should also be used, even if this is in an ambulance that is well equipped. Sometimes non-sitters and sitters may need to be transferred between hospitals as they should be cared for at a specialist (tertiary) centre that is equipped to look after them. The clinical team responsible for their long-term care should always be notified about the illness.

Further reading

- Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care
- Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics
- NICE (2019) Nusinersen for treating spinal muscular atrophy. Technology appraisal guidance [TA588]
- NICE (2016) Transition from children's to adults' services for young people using health or social care services [NG43]
- NICE technology appraisal: Risdiplam for treating spinal muscular atrophy in children and adults (publication expected July 2021)
- Care Quality Commission (2014) From the pond into the sea: Children's transition into adult health services

Organisations

- Muscular Dystrophy UK www.musculardystrophyuk.org
- SMA Reach UK www.smareachuk.org
- Spinal Muscular Atrophy UK www.smauk.org.uk
- Royal College of General Practitioners www.rcgp.org.uk

Glossary of abbreviations

BiPAP	Bilevel positive airway pressure
CT scan	Computerised tomography scan
EHP	Emergency healthcare plan
EAMS	Early Access to Medicines Scheme
EMG	Electromyogram
MAA	Managed access agreement
MDT	Multidisciplinary team
MRI	Magnetic resonance imaging scan
NICE	The National Institute for Health and Care Excellence
NIV	Non invasive ventilation
NG tube	Naso-gastric tube
NJ tube	Naso-jejunal tube
NM	Neuromuscular
OFC	Occipito-frontal circumference
OT	Occupational therapy
RSV vaccine	Respiratory syncytial virus vaccine
SCR	Shared-care record
SLA	Service level agreement
SMA	Spinal muscular atrophy

Patients may already be in the system and enter adult services through a transition pathway or referred in for assessment for one of the treatments now available for SMA

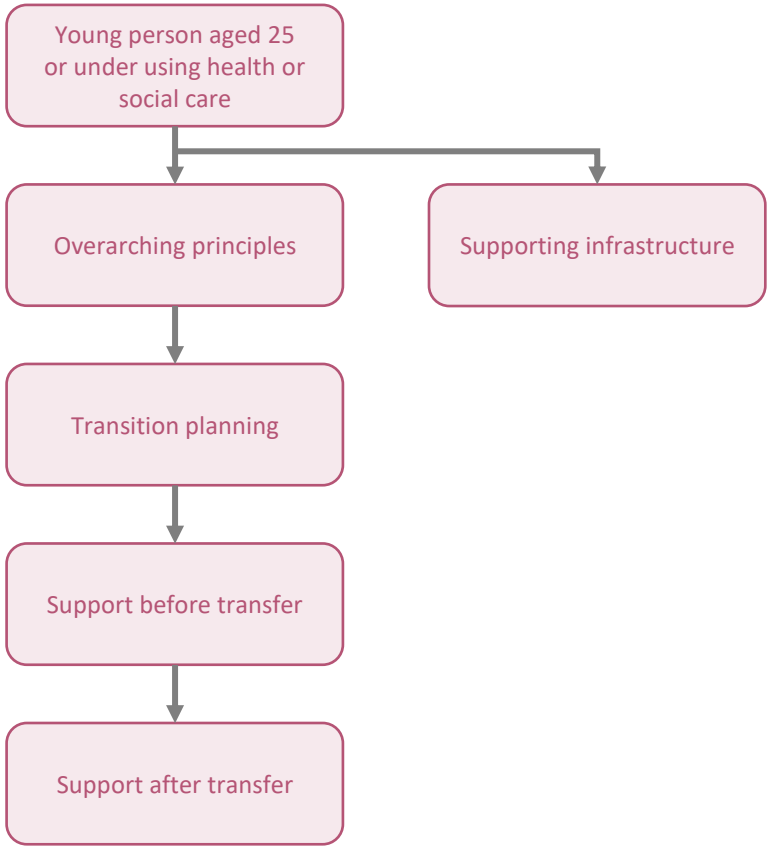
Transitioning from paediatric to adult care

In young people with chronic disabilities like SMA, the transition from paediatric to adult care is often difficult if structured and supportive transition programmes are not in place. The transition to adult care is often described as 'challenging and scary'.

- Learning to navigate a new and complex healthcare system.
- Differences in information provision and expectations.
- Engaging with unfamiliar specialists.
- Difficulty identifying and accessing specialists and multidisciplinary clinics.
- Difficulty accessing funding and equipment.
- Major resource gaps and lack of support navigating the system.

See also:

- Care Quality Commission (2014) From the pond into the sea: Children's transition into adult health services
- NICE (2016) Transition from children's to adults' services for young people using health or social care services [NG43]



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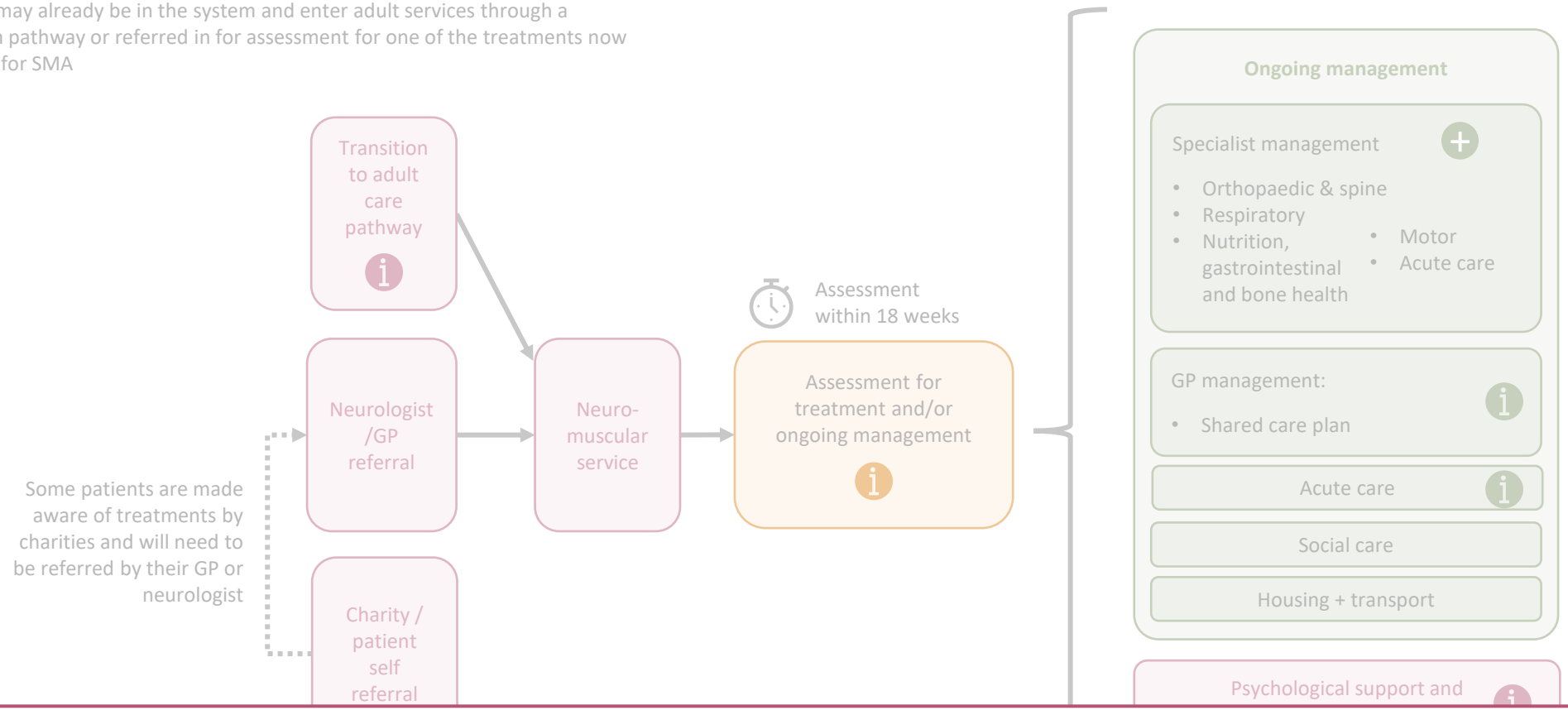


Psychological support and genetic counselling

- Psychological support for coming to terms with the diagnosis.
- What to expect from treatment and management.
- Genetic counselling, e.g. around family planning.

Patients may already be in the system and enter adult services through a transition pathway or referred in for assessment for one of the treatments now available for SMA

GP role

While the GP is not the primary point of contact for patients because they lack the necessary expertise, they can play a role in SMA care and the relationship is key in terms of delivering a quality service throughout a patient's life. Currently patients tend to bypass GPs altogether and go straight to the paediatric / neuromuscular specialist centre for help. However, with GP education and a comprehensive care plan, GPs can certainly be more engaged in supporting SMA patients with:

- Vitamin D prescription
- Importance of vaccinations including the specific types (e.g. RSV vaccine, although a change in national guidance is also needed here).
- Antibiotic prescriptions (GPs need training on the longer period recommend for these patients [10–14 days] and the low threshold for prescription for patients with frequent chest infections).

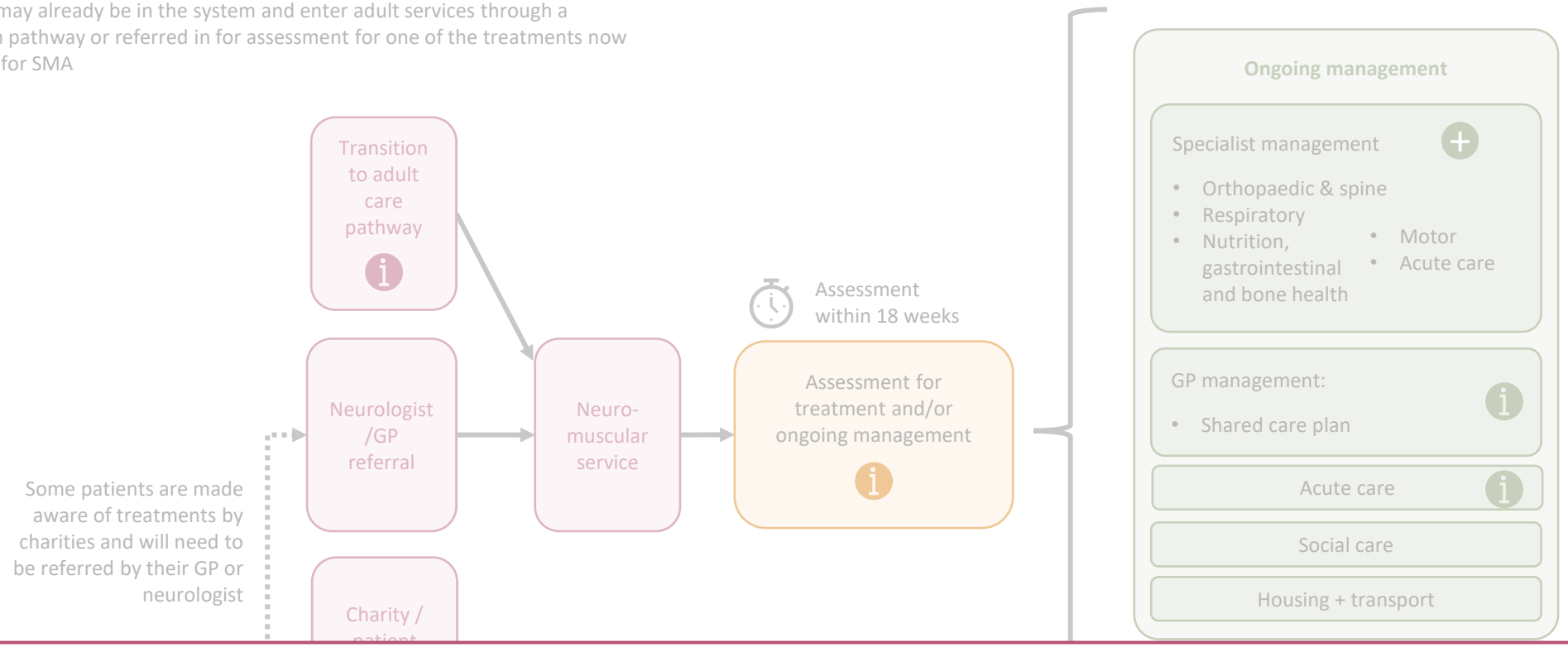
Each patient should have a shared care plan in place which indicates which symptoms they should contact their GP about (with accompanying advice for the GP) and which should be directed towards the paediatric/specialist centre.

Each patient should have a separate emergency care plan.

Community nurses can assist with: providing suction, be available to do swabs or take samples if required, NG replacement, and providing nebuliser machine.

Currently it appears that service level agreements (SLAs) are informal and shared-care records (SCRs) are not in place. This needs to change if there is to be effective working between the specialist centre and local teams.

Patients may already be in the system and enter adult services through a transition pathway or referred in for assessment for one of the treatments now available for SMA



Objectives for SMA service

1. Provide a specialist multidisciplinary neuromuscular service for diagnosis and ongoing management.
2. Initiate appropriate pharmacological and non-pharmacological treatments for patients with SMA.
3. Reduce morbidity and mortality due to SMA including reducing hospitalisation.
4. Ensure equity of access to specialised therapies.
5. Oversee all aspects of care that fall outside the expertise of local units.
6. At an individual level ensure the commissioning service is responsible for minimising disease impact in SMA.

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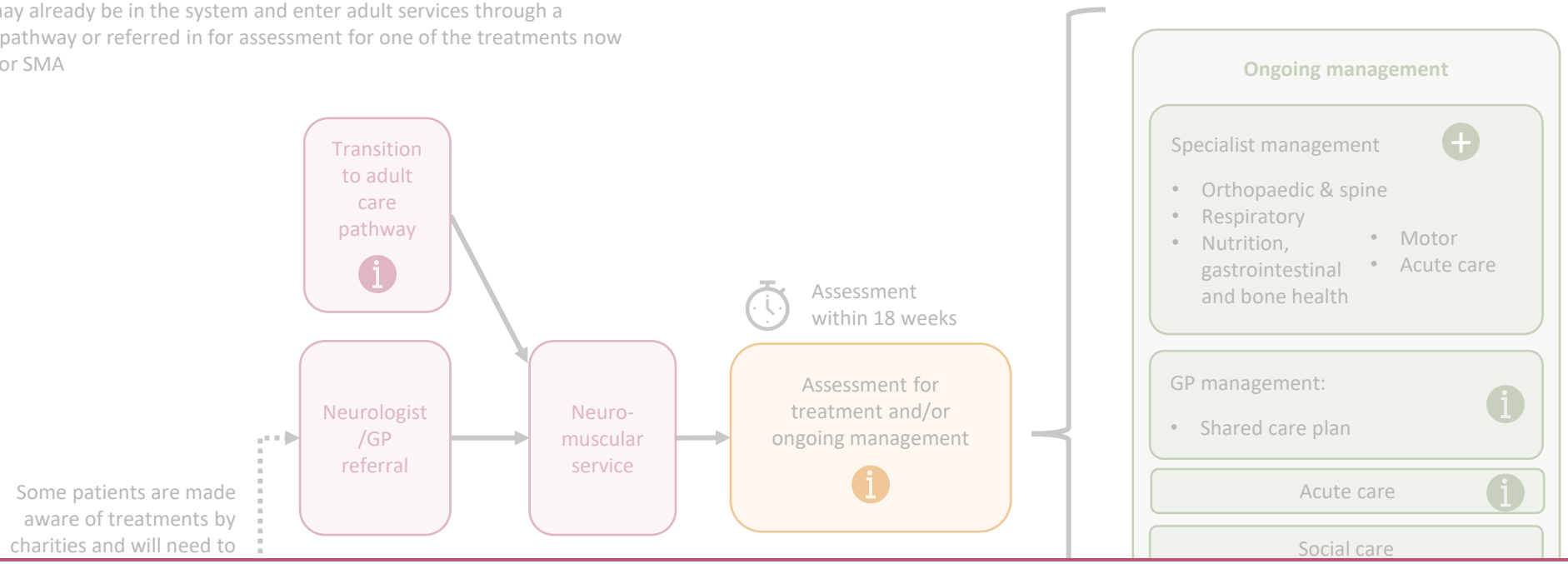


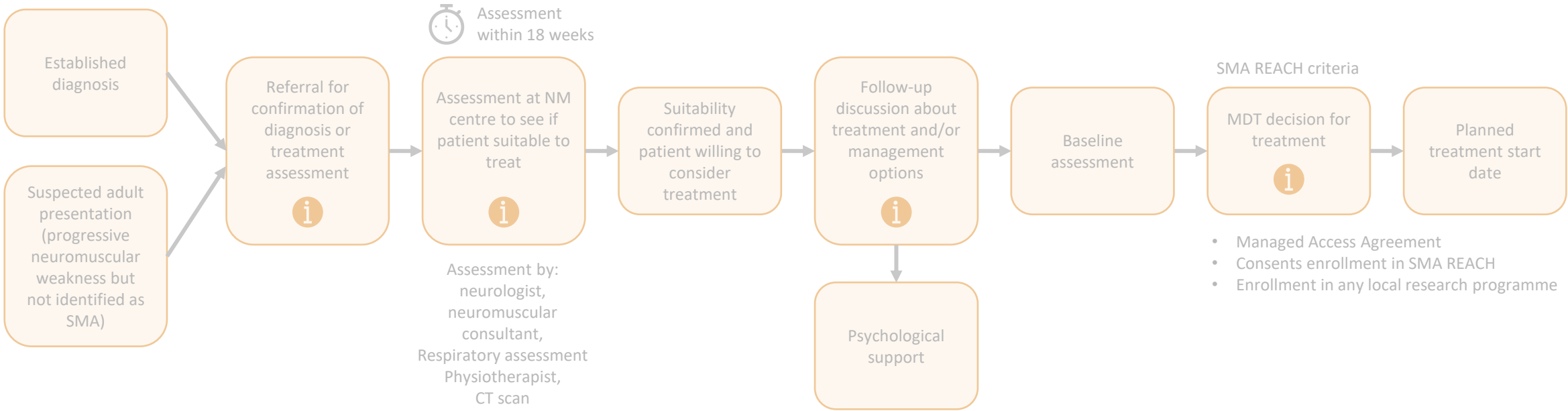
Audit points

- Timing of referrals:
  - Urgent referral within 1 day to 3 weeks depending on severity.
- Patient satisfaction questionnaire (local document) covering:
  - Patient made aware of identified timeframes.
  - Contact details at first consultation are made available to patient and their carers.
  - Patient received list of patient information at first consultation.
  - Patient referred for specialist medication (if appropriate) and received information leaflet about the medication detailing side effects and instructions about monitoring.

Patients may already be in the system and enter adult services through a transition pathway or referred in for assessment for one of the treatments now available for SMA

Key performance indicators

- Identified benefit to patients.
    - Equity of access.
    - Appropriate referral pathway based on evidenced-based clinical assessment.
    - Access to the specialist team.
    - Patient has understanding of prognosis, available treatment and outcomes.
- Benefit to health professionals and organisation.
    - Appropriate referral and prioritisation of patient care achieved.
    - Standardisation of assessment and referral process.
    - Relevant investigations and patient information available, reducing delayed in patient journey later in pathway.
    - SMA MDT coordinator to liaise and respond directly with health professional and patient/carer regarding referral enquiry, waiting list management and liaise with the appropriate SMA team member to provide supportive information.
    - Cost and time effective management.
    - Clearly defined diagnostic process to follow.
    - Appropriate use of professional expertise and designated discussion time.
    - Clearly defined roles and responsibilities of the health professionals involved in the pathway.



- Adult onset ambulatory (Type IV). Symptom onset aged 19 or older. The patient takes at least five steps independently in the upright position with the back straight. One leg moves forward while the

Referral

There may be a video consultation ‘pre-screen’ at this point to establish when the patient was last seen, discuss potential genetic testing to confirm diagnosis and the implications of having treatment.

Assessment
within 18 weeks

Established

SMA REACH criteria

Assessment

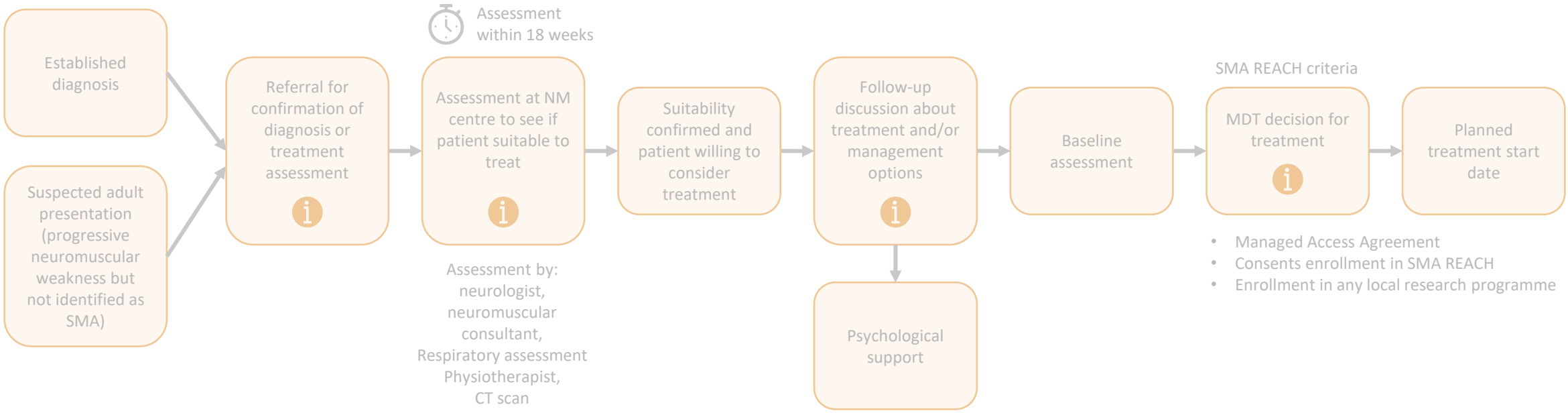
This may require a genetic test to confirm diagnosis if the patient has not been seen for some time. SMA Type 4, the more common form of adult onset SMA, is most often diagnosed in early adulthood. There are other forms of adult onset SMA with different causes. The impact of adult onset SMA varies greatly between individuals.

Getting a diagnosis of SMA Type 4 or other form of adult onset SMA can take time as the symptoms of SMA can seem similar to the symptoms of other neuromuscular conditions. Waiting for appointments, test results and a diagnosis can be very stressful. If you're concerned about your symptoms, have had some tests but have not been referred to a consultant neurologist, you might want to request a referral from your GP.

Possible tests are:

- An EMG which shows if the nerve supply is diminished.
- A muscle biopsy which shows if there is any reduction in muscle cells.
- An MRI scan which produces detailed images of the inside of the body.
- A CT scan which is another way of producing detailed images of the inside of the body.
- A range of blood tests, including a blood sample for DNA testing and baseline blood profile.
- Physiotherapy and respiratory assessment may also be required if the patient has not had a review for some time to ensure there are no comorbidities that might impact treatment.

Even with all these tests it's not always straightforward to say exactly what neuromuscular condition an adult has, though a specific diagnosis of SMA Type 4 can be made following DNA testing.



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Follow-up discussions about treatment and/or management options

Discussion about the full implications of intrathecal treatment, the requirement to have this administered in hospital (may necessitate spinal imaging by interventional radiology to assess access) and the impact on the patient's current routine.

SMA REACH

SMA REACH UK is a two year study funded by a grant from SMA UK to Great Ormond Street Children's Charity. The project is a new initiative in collaboration with existing UK SMA registries – the UK SMA Patient Registry and SMArtNet Clinical Network UK sponsored by Muscular Dystrophy UK and Spinal Muscular Atrophy Support UK (formerly The Jennifer Trust). The SMA REACH UK project is led by the study team based at the Dubowitz Neuromuscular Centre, UCL.

The primary aim is to establish the first national clinical and research network: SMA REACH UK – SMA Research and Clinical Hub UK – to promote a national agreement on clinical and physiotherapy assessment and standards of care. The plan is to design, pilot and expand an electronic database created to streamline the collection of data for patients with SMA. This UK SMA database would be a unique infrastructure started at GOSH and Newcastle which will be accessible to specialist centres across the UK who treat patients with SMA.

The secondary aim of the project is to utilise the SMA REACH UK database as a longitudinal data store where information can be audited and reviewed. This will provide clinicians and researchers a rich resource of available information on a large collection of SMA patients. SMA Reach are collaborating with the Catholic University Rome, an international centre of excellence in SMA research and treatment, with the shared goal to facilitate translational research for this common neuromuscular disease in preparation to design national and international clinical trials. Once the system is finalised, additional national sites that have a history of successful SMA enrolment will be invited to participate and collect high quality longitudinal data.

This work will be an invaluable tool for the centres likely to be involved in upcoming SMA multicentre randomised clinical trials in SMA type I, II and III and ensure that the functional scales used are suitable and clinically relevant for future trials.

SMA REACH is the UK's SMA research and clinical hub UK focused on improving standards of care and translational research. It aims to establish national agreement on medical and physiotherapy assessments, and standards of care for patients with SMA in preparation for future clinical trials in the UK.

Read more on the SMA REACH website

Anaesthetics

If the patient requires an anaesthetic the GP should alert the hospital if this is an elective admission to ensure the team in charge undertake:

- A review of the heart by a cardiologist (only if there is known to be a pre-existing problem)
- A full assessment before any anaesthetics are used. This may include a sleep study and involvement of a dietitian.
- Assessment of difficulties in intubating that may be caused by:
    - Tightening of the jaw
    - Limited neck mobility
    - Difficulties in positioning
- Monitoring blood carbon dioxide and oxygen at all times, whatever anaesthetic method is used.
- Anticipating any other possible needs such as use of NIV and other breathing interventions.
- Medication for pain management may be needed after an operation.

Considerations of local anaesthesia or regional analgesia; as a general rule anaesthesia does bring challenges.

Anaesthetics may be used for planned surgery as well as for other reasons, such as administering new drug treatments.

Comply with reporting data, assess patient

Early Access to Medicines Scheme (EAMS)

The EAMS programme helps to give people with life threatening or seriously debilitating conditions early access to new medicines that do not have a marketing authorisation but where there is a clear unmet medical need. By promoting early engagement between pharmaceutical companies, and the Accelerated Access Consortium (AAC) partners including MHRA, NICE, NHS England and NHS Improvement, EAMS also helps to create a smoother route to market for new treatments. Since launch of the scheme in 2014 over 1,200 people with conditions ranging from cancer to Duchenne Muscular Dystrophy, from across the UK, have benefited from early access to new treatments through EAMS. Risdiplam is currently going through a NICE Health Technology Appraisal (HTA) and the expected publication date is in 2021. Patients currently accessing the drug now do so under the EAMS programme.

EAMS has been granted for risdiplam for type 1 or type 2 SMA who are not suitable for authorised treatments. EAMS scheme is now closed to new patients following marketing authorisation from the MHRA (May 2021). Existing patients participating in the EAMS can continue to be treated until reimbursement.

- https://www.gov.uk/guidance/apply-for-the-early-access-to-medicines-scheme-eams
- https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/918768/Risdiplam_Treatment_protocol_Information_for_patients.pdf

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Homecare

Homecare can be used to supply the drug. The homecare company can reconstitute the drug powder and deliver the resultant oral solution to the patient using cold chain storage. Risdiplam is supplied via the homecare pharmacy or specialist treatment centre. If risdiplam is supplied by the specialist treatment centre, they will:

- Provide sufficient risdiplam to take at home/outside of hospital setting.
- Provide instructions on how bottles can be transported home
- Provide instructions on how to store and take the medication.
- Supply oral syringes for medicine administration.

If risdiplam is supplied by homecare pharmacy, they will:

- Deliver sufficient risdiplam to the home or previously agreed address.

The patient is also provided with an “Instructions for use for patients/parents and carers” booklet. This booklet gives detailed instructions on how to prepare the dose volume with the re-usable oral syringes provided, and take the medicine either: by mouth, through a gastrotomy tube, or through a nasogastric tube.