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SMA  
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# Spinal Muscular Atrophy: Paediatric Care Pathway

ENTER





## 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 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 infants may wait too long for a referral and diagnosis which can have major ramifications for their symptom management, quality of life and life expectancy. It is essential that the complexity of SMA care is understood and we hope this integrated care pathway will help to unravel the SMA journey and aid improvements and streamlining the care delivered.

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

- The child's and their family's 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 atrophy (SMA) is 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:

### 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

## 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.



## Every day counts

Early diagnosis is very important as there are now available effective treatments for all SMA types but it is crucial for SMA type I and II.

Diagnosing Spinal Muscular Atrophy:  
**Every Day Counts**

This activity has been supported by sponsorship from Biogen Idec Ltd.  
The sponsor has had no control over the educational content of this activity.

Watch the video at: <https://vimeo.com/546458032/532f462d1d>



## Acknowledgements

Our thanks to the following individuals who contributed to the development of the integrated care pathway:

- Sian Ball, Neuromuscular Care Advisor, Sheffield Children's NHS Foundation Trust
- Lisa Edel, Respiratory Neuromuscular Physiotherapist, Great Ormond Street Hospital for Children NHS Foundation Trust
- Dr Vasantha Gowda, Consultant Paediatric Neurologist, Guy's and St Thomas' NHS Foundation Trust
- Janet Johnson, Neuromuscular Specialist Practitioner, Manchester University NHS Foundation Trust
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- Sandya Tirupathi, Consultant Paediatric Neurologist, Belfast Health and Social Care Trust
- Dr Elizabeth Wraige, Consultant Paediatric Neurologist, Guy's and St Thomas' NHS Foundation Trust

### Facilitator

- Sue Thomas, Independent Healthcare Consultant

### Pathway design

- Sarah Mehta, Medical Writer

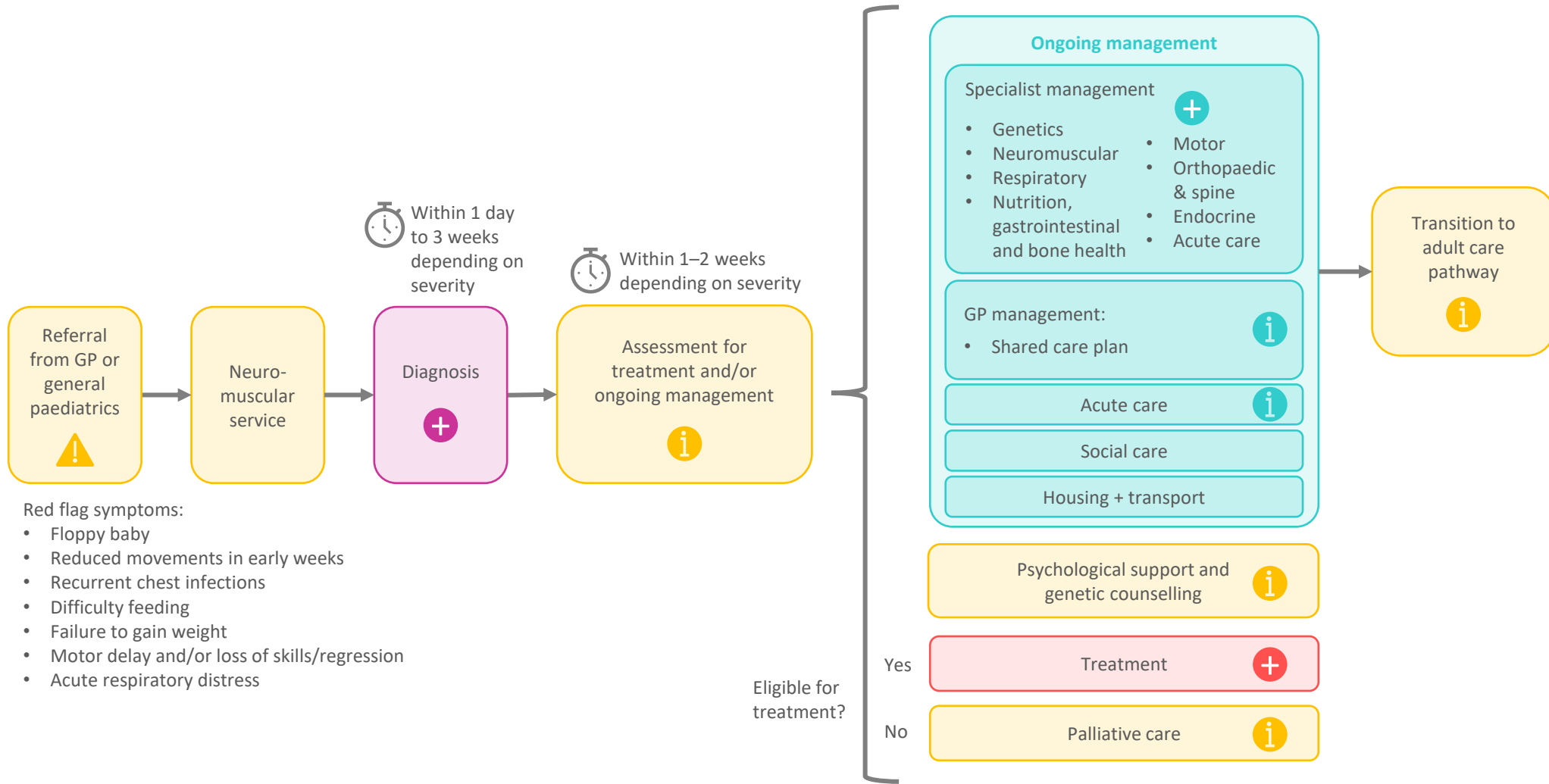
## Contact us

Please contact us with your comments and feedback at: [info@neurologyacademy.org](mailto:info@neurologyacademy.org)

Document due for review 1 September 2021.

Centres involved in the development of this pathway:

- Barts NHS Health Trust
- Belfast Health and Social Care Trust
- Great Ormond Street Hospital for Children NHS Foundation Trust
- Guy's and St Thomas' NHS Foundation Trust
- Manchester University NHS Foundation Trust
- Nottingham University Hospitals NHS Trust
- Oxford University Hospitals NHS Foundation Trust
- Sheffield Children's NHS Foundation Trust
- University Hospitals Birmingham NHS Foundation Trust

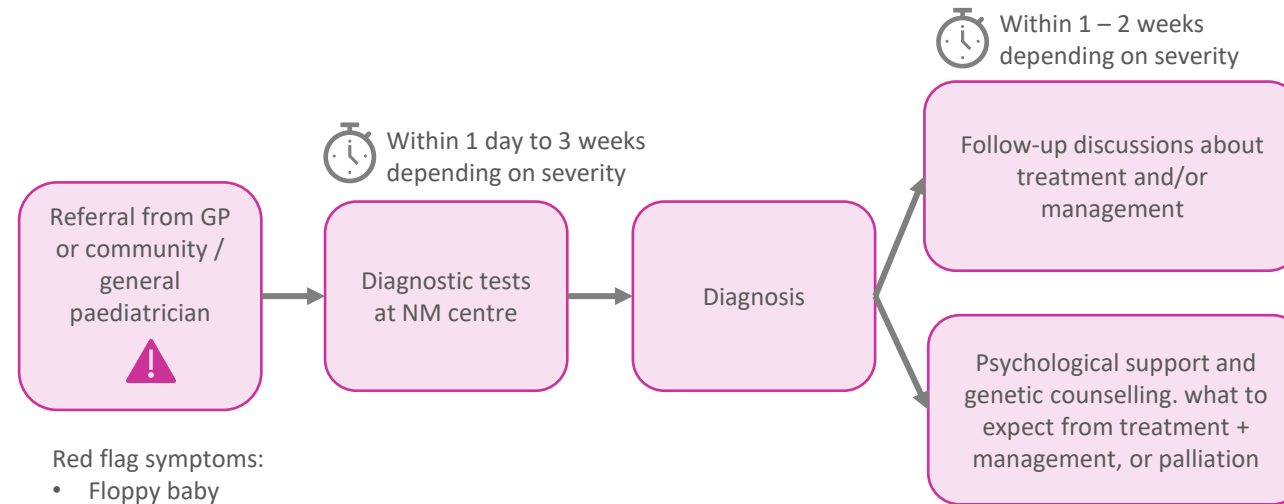


- Red flag symptoms:
- Floppy baby
  - Reduced movements in early weeks
  - Recurrent chest infections
  - Difficulty feeding
  - Failure to gain weight
  - Motor delay and/or loss of skills/regression
  - Acute respiratory distress

Objectives

Audit points

KPIs



Red flag symptoms:

- Floppy baby
- Reduced movements in early weeks
- Recurrent chest infections
- Difficulty feeding
- Failure to gain weight
- Motor delay and/or loss of skills/regression
- Acute respiratory distress

Early diagnosis and treatment avoids emergency care appointments.

The Expert Working Group recommended that infants are reviewed as soon as possible. This will depend on local wait times but should be between one day and a maximum of three weeks.



## 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 nusinersen 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 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 those patients who may not be able to tolerate intrathecal injections or are not eligible for nusinersen.

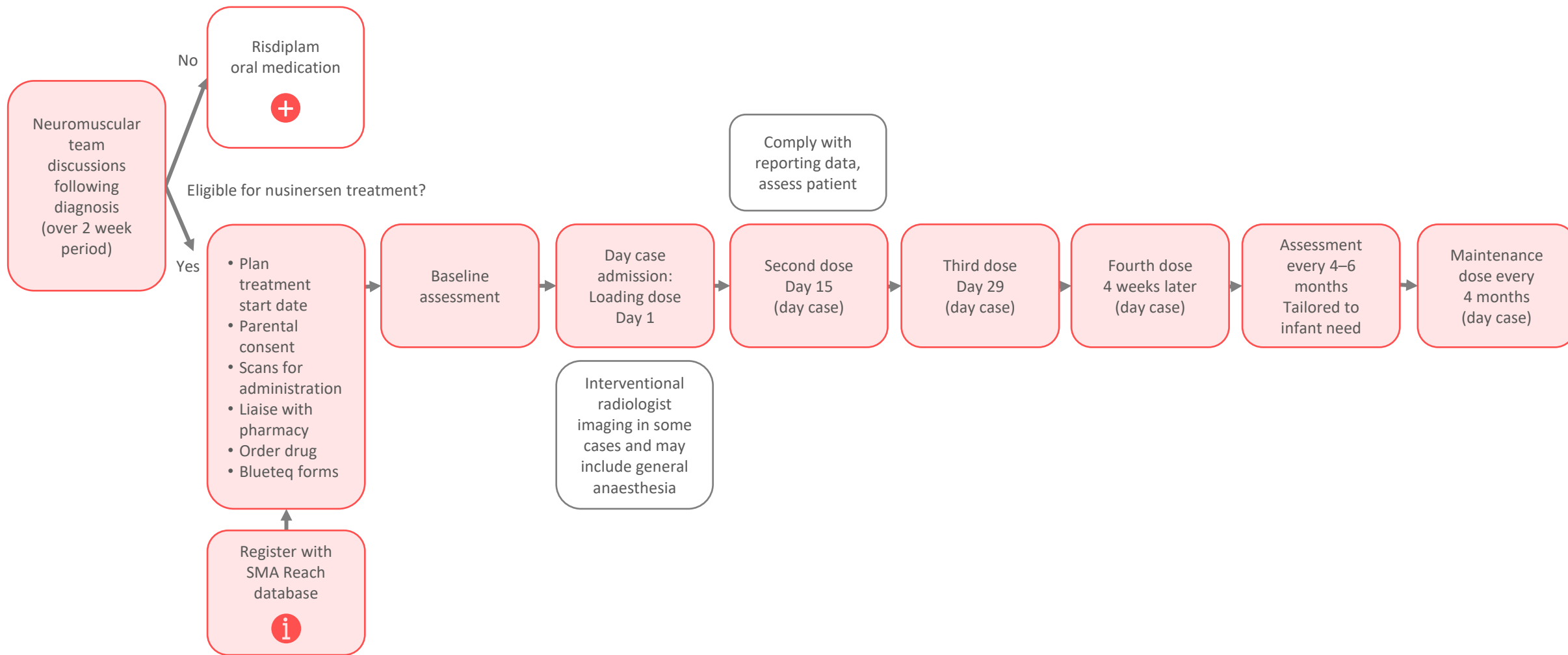
- [NICE technology appraisal: Risdiplam for treating spinal muscular atrophy in children and adults \(publication expected July 2021\)](#)
- [Further information about EAMS](#)

## Onasemnogene abeparvovec (Zolgensma)

This gene therapy treatment, delivered by IV injection, is designed to address the genetic root cause of SMA and limit the progression of the condition by replacing the faulty or missing SMN1 gene. NICE published draft guidance on 8 March 2021 recommending this £1.79 million treatment for babies aged up to 12 months with SMA I who would normally have a life expectancy of less than 2 years. The draft guidance also recommends the treatment for babies with SMA before they develop symptoms as part of an MAA while further data is collected. Despite the high cost of the treatment the recommendations are made because the evidence is of exceptional benefit to young babies potentially allowing them to reach normal childhood development milestones. Provision of each treatment will be discussed by a national multidisciplinary team.

- [Onasemnogene abeparvovec for treating spinal muscular atrophy type 1 \[ID1473\]. In development \[GID-HST10026\]](#)







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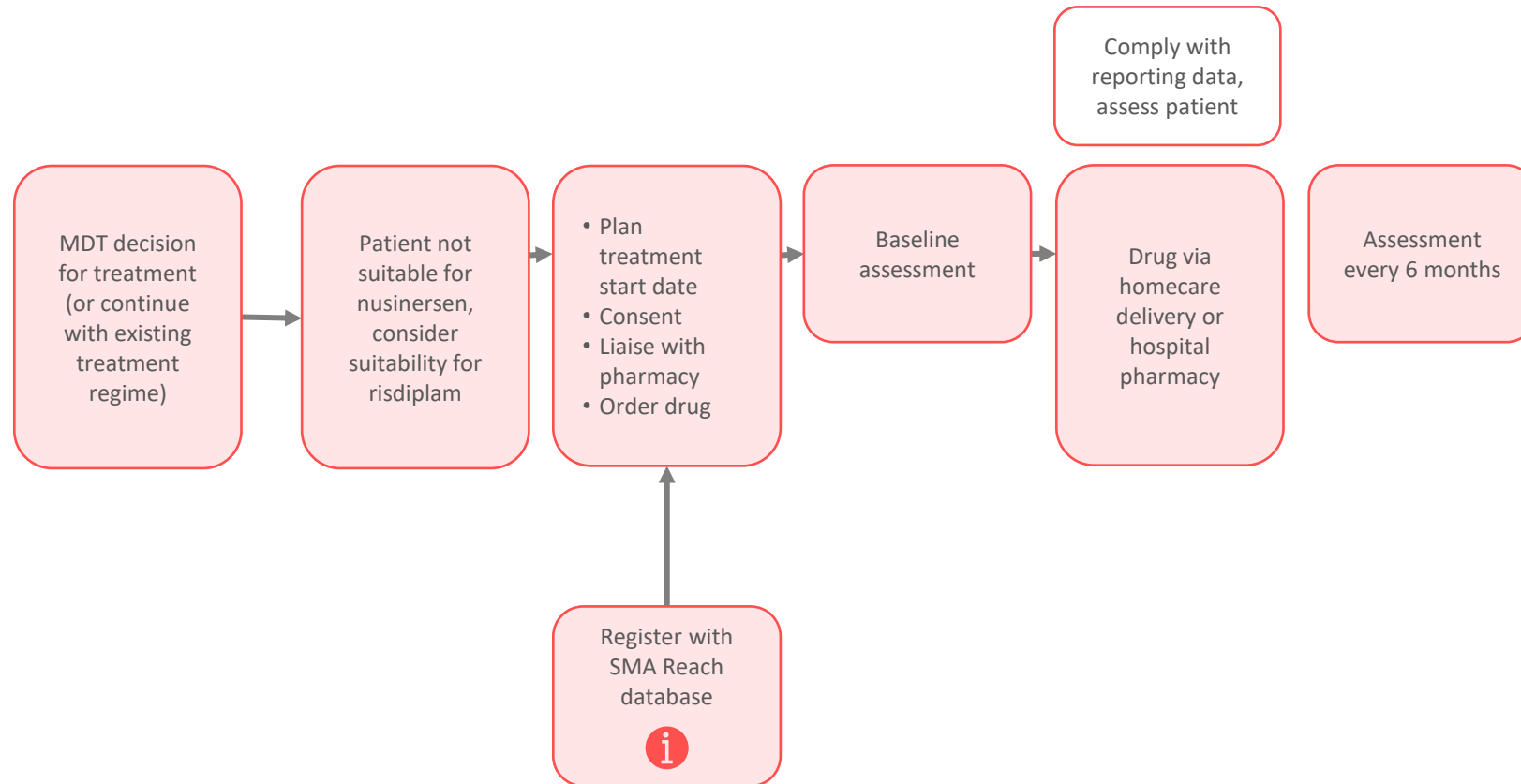
Glossary

Overview

Nusinersen

Risdiplam

Onasemnogene abeparvovec



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## Onasemnogene abeparvovec

At the time of publishing this pathway a new global clinical trial is planned that will be investigating the safety, tolerability and efficacy of intravenous onasemnogene abeparvovec in children who have SMA and weigh between 8.5 and 21 kg.

This global trial has an estimated start date of 30 September 2021 and aims to recruit 24 participants in total across sites in Europe, North America, Australia and Taiwan, and will follow patients for a period of 12 months. We understand Novartis is currently exploring the potential of setting up sites in the UK.

NHS England News, 8 March 2021:

[NHS England strikes deal on life-saving gene-therapy drug that can help babies with rare genetic disease move and walk](#)

Administration: Onasemnogene abeparvovec uses harmless, genetically engineered viruses to increase SMN protein levels. Once the virus is introduced into a person, it is able to travel around the body and get to a variety of different cells to help restore some of the SMN protein that is missing in SMA. These viruses cannot produce copies of themselves, and are therefore unable to be passed from person to person. Currently delivery is via intravenous injection. This method has been tested in clinical trials with children up to two years of age.

[Onasemnogene abeparvovec for treating spinal muscular atrophy type 1 \[ID1473\]. In development \[GID-HST10026\]](#)



## Neuromuscular management

A multidisciplinary team approach is the key element in management of SMA infants and children and the different aspects of care should not be dealt with in isolation but as part of an MDT approach taking into consideration all aspects of daily living including school and social activities. 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 infants and children.

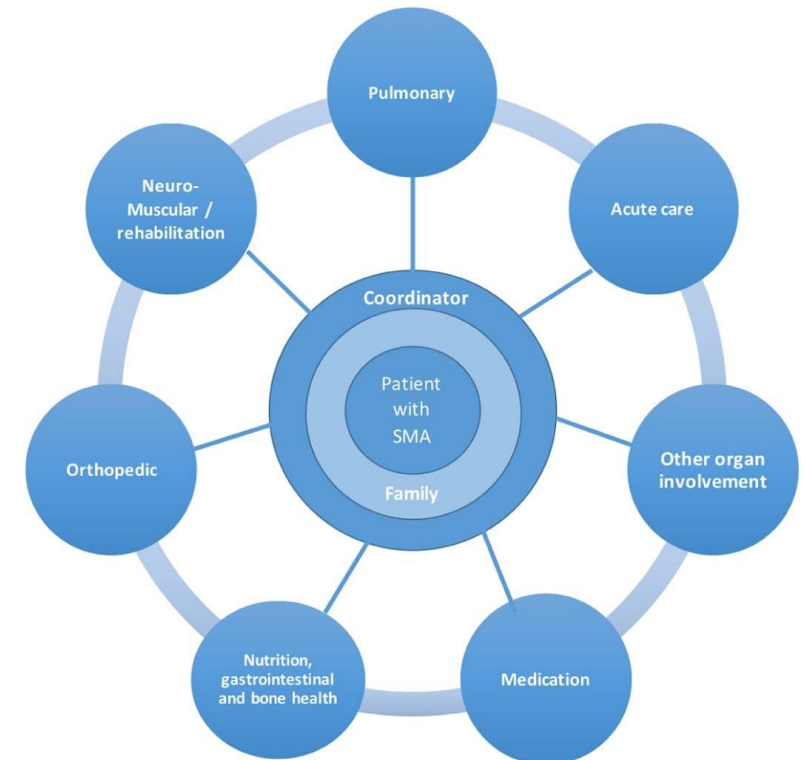
The key to all management is a baseline assessment and regular review of the child's ability (see [SMA outcome measures](#) and [measures evaluated in clinical trials](#)) . Any improvement or maintenance is good.

Rehabilitation: physiotherapy, occupational therapy, speech and language therapy:

- Contracture management
- Movement abilitation
- Oromotor abilitation
- Cognitive stimulation.

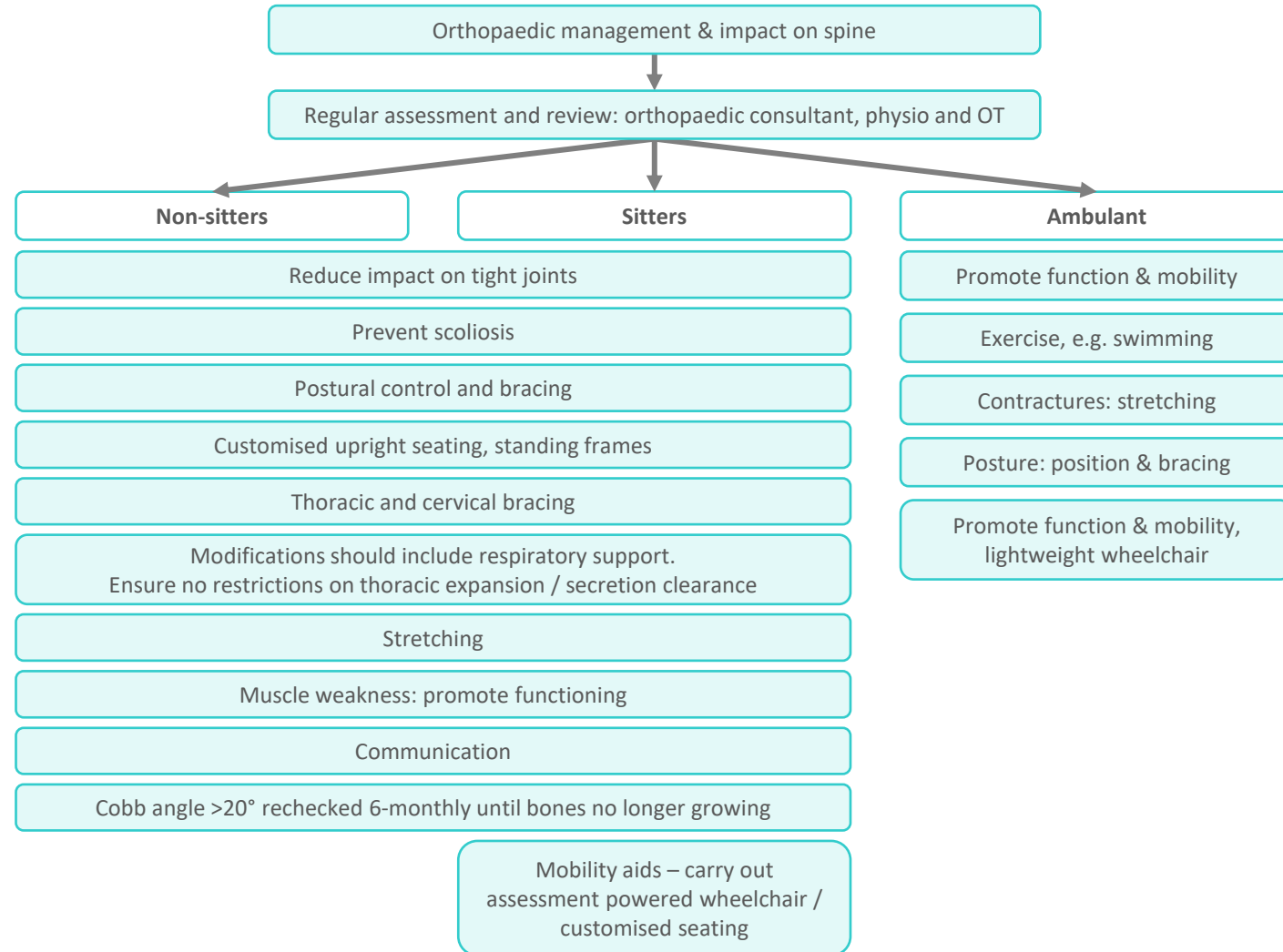
Respiratory rehabilitation and management:

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



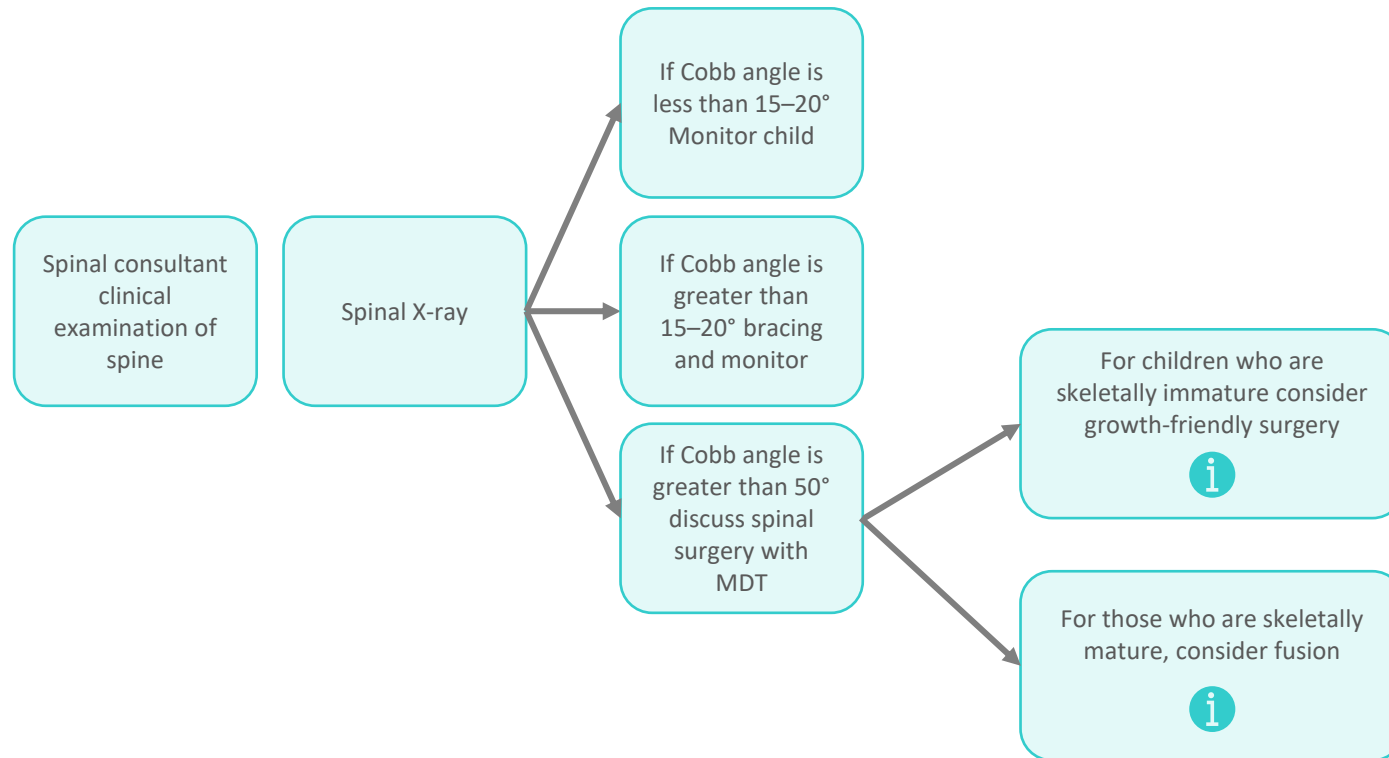
Multidisciplinary approach.

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





## Clinical management route a clinician will take when they suspect a person has curvature of the spine



## Recommended age of spinal intervention:

The standard of care recommendations to medical teams vary depending on age and are as follows:

- **Under 4 years:**  
In general, spinal surgery should be delayed until after 4 years of age.
- **Age 4 – 10 years:**  
As children are skeletally immature, growth-friendly spinal surgery is recommended.
- **Age 10 – 12 years:**  
At this stage children are transitioning to skeletal maturity. If surgery is needed, the type will depend on the child's skeletal maturity and how much more their spine is likely to grow.
- **Age over 12 years:**  
This is when children are skeletally mature. If surgery is needed, spinal fusion surgery is recommended.



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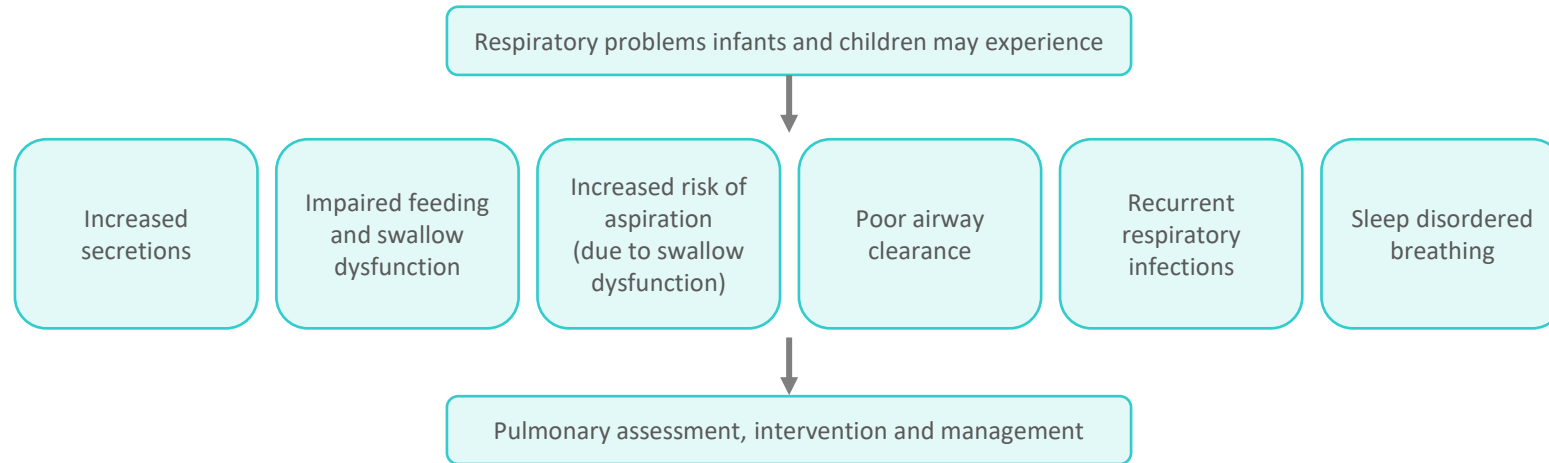
Orthopaedic & spine

Respiratory

Nutrition, GI & bone health

Motor

Acute care



Next »

**Non-sitters****Assessment**

- Clinic visits every 3–6 months.
- Monitor SpO<sub>2</sub> and CO<sub>2</sub>.
- Sleep study to confirm sleep disordered breathing/respiratory failure and indicate NIV.
- Assess for reflux.

**Intervention:**

- Proactive.
- Ensure full MDT.

**Airway clearance**

- Clinical assessment of PCF.
- Initiate individualised chest physiotherapy (this may include manual CPT, effective oral/nasal suctioning and cough assist).

**Ventilation**

- Use bilevel ventilation in all symptomatic infants.
- Ensure correct interface.
- Avoid CPAP.

**Medications**

- Consider mucolytics if secretions are problematic.
- Glycopyrrolate for salivary management.
- Ensure RSV vaccine up to 24 months and influenza vaccine annually during winter months after 6 months of age.
- Pneumococcal vaccine with addition of Pneumovax(R) from 2 years of age.

**Sitters****Assessment**

- Clinic visits every 6 months.
- Sleep study to assess for nocturnal hypoventilation and indicate NIV.
- Assess for reflux.
- Spirometry (depending on age/cooperation.).

**Intervention:**

- Proactive.
- Ensure full MDT.

**Airway clearance**

- Clinical assessment of PCF.
- Initiate individualised chest physiotherapy (this may include manual CPT, effective oral/nasal suctioning and cough assist).

**Ventilation**

- Use bilevel ventilation in all symptomatic infants.
- Ensure correct interface.
- Avoid CPAP.

**Medications**

- Ensure RSV vaccine up to 24 months and influenza vaccine annually during winter months after 6 months of age.
- Pneumococcal vaccine with addition of Pneumovax(R) from 2 years of age.

**Ambulant****Referral for respiratory team if:**

- Evidence of weak cough.
- Recurrent infections.
- Signs/symptoms of nocturnal hypoventilation.

**Medications**

- Annual influenza vaccine.
- Pneumococcal vaccine with addition of Pneumovax(R) from 2 years of age.

NB. If bilevel ventilation is not available in a local hospital during an acute respiratory episode, stabilise on CPAP/Optiflow and monitor O<sub>2</sub>/CO<sub>2</sub>. It is then vital the patient moves to a tertiary centre.





### The respiratory pathway at Great Ormond Street

**Referral to respiratory neuromuscular physiotherapist**  
Set up with respiratory physiotherapy management plan

**Ongoing respiratory physiotherapy**  
Positioning, manual techniques, suction, nebulisers, MIE

**Referral to community care**

**Respiratory physiotherapy clinic follow up / telephone support**

**Diagnosis meeting**  
NMD team and palliative care team

**Referral to NIV team**  
NMS team and palliative care team liaise with NIV team

**Meeting on sleep unit/respiratory clinic with parents, NIV clinical nurse specialist & respiratory consultant**  
Discussion about respiratory management (including pros and cons of NIV)

**Baseline sleep study**  
Arranged according to clinical assessment and parental consent to trial of NIV if indicated

**Acclimatisation**

**Elective admission for initiation**  
Issue of NIV equipment and training session for parents. Involvement of community team and palliative care team.

**Respiratory clinic follow up**  
Follow-up sleep study for monitoring of ventilation. Home visit with palliative care team. Telephone support.

### Great Ormond Street criteria for NIV initiation

≥1 main criteria +/- supportive criteria

Note: supportive criteria alone are not sufficient indications for initiation of NIV

#### Main criteria

1. Infective exacerbation (acute)
2. Recurrent chest infections
3. Baseline increased work of breathing/dyspnoea
4. Documented respiratory failure – either chronic (as shown on sleep studies) or acute

#### Supportive criteria

1. Poor weight gain in spite of optimised feeding
2. Chest deformity

Example pathway from Great Ormond Street Hospital



## Key messages

- Safe swallowing is one of the most important aspects of care as children with a weak swallow are at risk of inhaling (aspirating) their feed which can cause choking and chest (respiratory) infections.
- Standards of Care recommend that a dietician reviews feeding and diet every 3–6 months for younger children and annually for older children.
- If swallowing becomes unsafe, or if the child is not gaining enough 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.



### Non sitters

Assessment safe swallowing – bulbar dysfunction can result in aspiration and pulmonary infections.

Oral feeding may be limited.

Interventions - failed swallow study NG or NJ tube.

Adequate hydration and electrolyte balance important during illness.

Video Fluoroscopic swallow study shortly after diagnosis and when suggested by clinical signs suggestive of dysphagia (weak suck, fatigue, pneumonias). Difficulties with feeding (pocketing, jaw contractures, increased feeding time). Nutritional analysis of food records/feeding regimen Longitudinal anthropometrics, acute care monitoring, 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 3–6 monthly for younger children and annually for older children. NB Evaluation essential important for those on specialised diets.

### Sitters

Assessment of symptoms of dysphagia/aspiration/difficulties with feeding. Video fuoroscopic swallow study if clinical signs suggestive of dysphagia. Nutritional analysis of food records/feeding regimen Longitudinal anthropometrics (height, weight, OFC) Nutrition labs may be indicated. Acute care monitoring Glucose metabolism labs 25 Hydroxy-vitamin D labs, body composition and bone density (DXA)

Constipation management – evaluation of fluid and fibre intake recommended for constipation

Dietician evaluation shortly after diagnosis and for concerns of under/over nutrition. Possibility of obesity greater than the 25th percentile. Optimal care evaluation by a dietitian 3–6 monthly for younger children and annually for older children. NB Evaluation important for specialised diets.

### Ambulant

Dietician assessment

- See dietitian if over/under nutrition
- Nutritional analysis/monitoring if underweight or overweight
- Longitudinal anthropometrics (height, weight, OFC). Glucose metabolism labs 25 Hydroxy-vitamin D labs

Bone health important

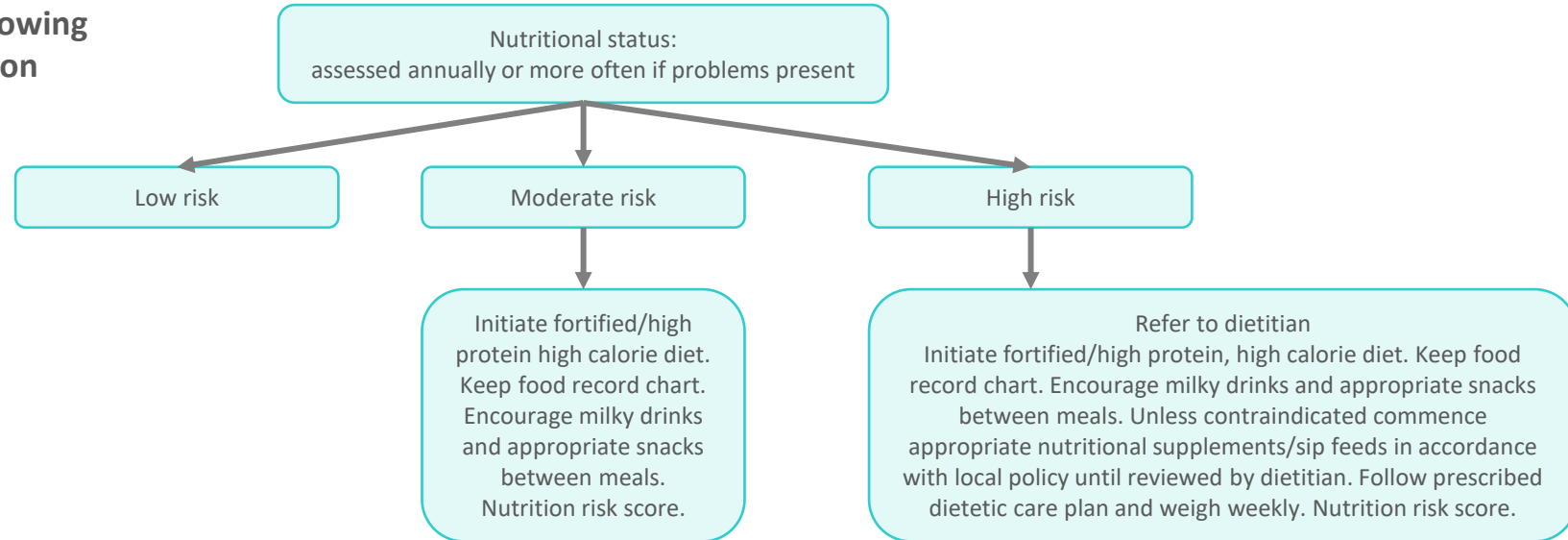
- High incidence of osteopenia and fractures
- DEXA scan

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

NB. Children with SMA are at risk of hypoglycaemia; following period of vomiting or fasting ensure sufficient fluids to prevent ketosis.



### Nutritional assessment, swallowing and gastrointestinal dysfunction and intervention



**Monitor weight, height and BMI**  
Children 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 children 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**  
Children 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.



## Physiotherapy and rehabilitation

### Non sitters

Physiotherapy and rehabilitation aims to reduce impact on tight joints, optimise function and help an infant/child tolerate different positions, lying or sitting with assistance. Regular assessment from physiotherapist and 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.
  - Serial casting which puts the limb into a series of plaster slowly correcting the position with each re-casting.
  - 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 infant/child 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:** 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. Beneficial to carry out an assessment before the age of 2. 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.

### Ambulant

Involvement of physiotherapy and occupational therapy.

The overall aim if to promote maximum mobility range of movement and as much independence as possible with day-to-day activities:

- Work on flexibility strength endurance and balance.
- Stretching.
- Positioning.
- Mobility and exercise.
- Lightweight manual wheelchairs or ones with power-assisted wheels useful as may be tiring to talk.
- Powered wheelchairs or scooters for long distance.

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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 child 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 parents, child 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.
- Nutritional and fluids needed.
- Techniques used for clearing secretion.
- When and which antibiotics should be given.
- Action agreed if resuscitation is required.
- Individual wishes, of the parents (for children) or child.



## 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 \[TA588\]](#)
- [Care Quality Commission \(2014\) From the pond into the sea: Children's transition into adult health services](#)

## Organisations

- Muscular Dystrophy UK [www.muscular dystrophyuk.org](http://www.muscular dystrophyuk.org)
- SMA Reach UK [www.smareachuk.org](http://www.smareachuk.org)
- Spinal Muscular Atrophy UK [www.smauk.org.uk](http://www.smauk.org.uk)
- Royal College of General Practitioners [www.rcgp.org.uk](http://www.rcgp.org.uk)



## Glossary of abbreviations

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





Within 1 day

Ongoing management

Specialist management

- Genetics
- Neuromuscular
- Respiratory
- Nutrition, gastrointestinal
- Motor
- Orthopaedic & spine
- Endocrine

Transition to



**Suspect SMA: red flags for GPs and health visitors**

Red flag symptoms that the GP, paediatrician, or intensive care should look for:

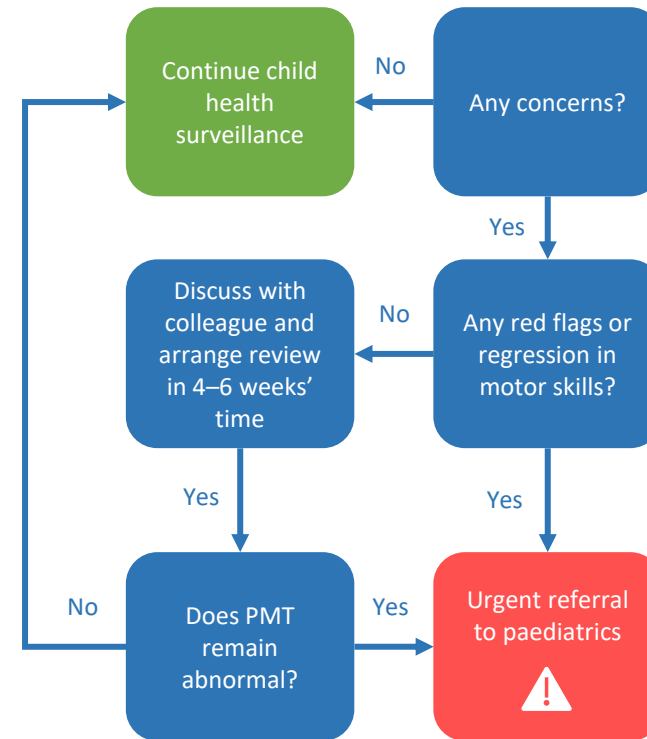
- Floppy baby
- Reduced movements in early weeks
- Recurrent chest infections
- Difficulty feeding
- Failure to gain weight
- Motor delay or loss/regression of skills
- Acute respiratory distress.

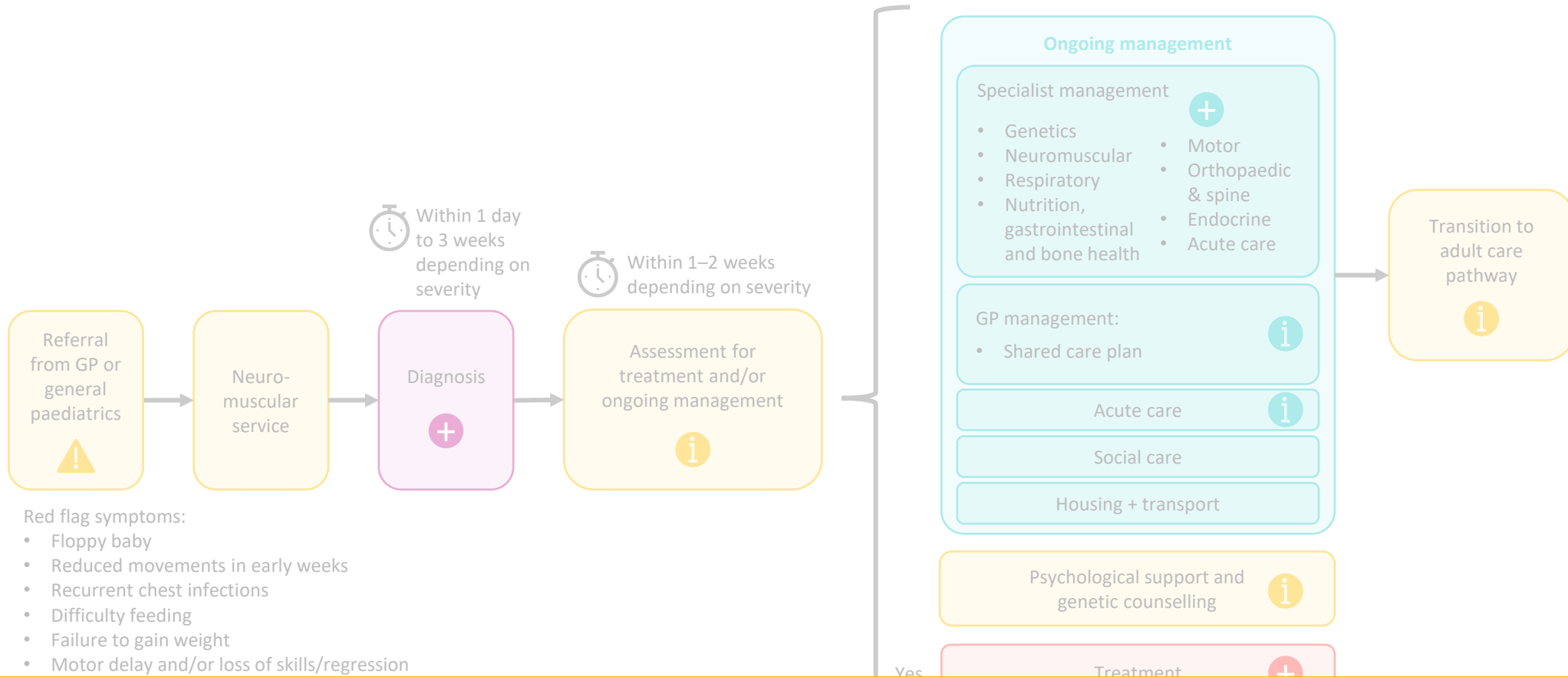
**Urgent referral**

Red flag symptoms that the GP, paediatrician, or intensive care should look for:

- Infants with acute respiratory distress requiring respiratory support.

Source: Recognising Neuromuscular Disorders – a practical approach eLearning module [www.rcgp.org.uk](http://www.rcgp.org.uk)

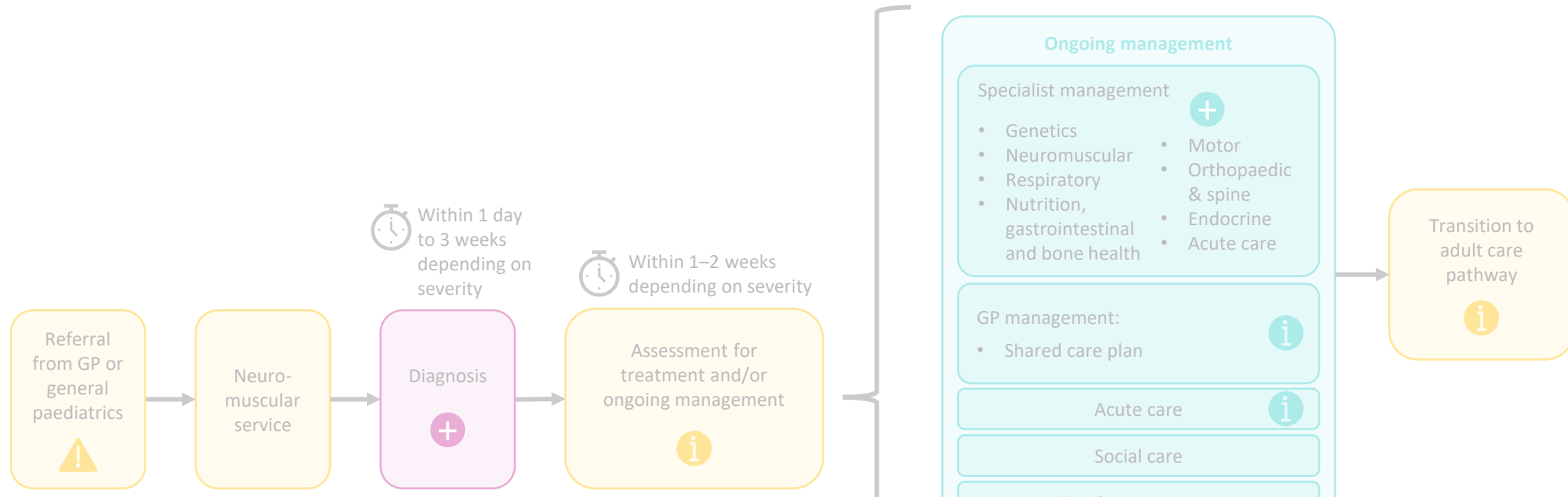




### Availability of treatment

Education of the family and GP is important to understand there are now treatments available.





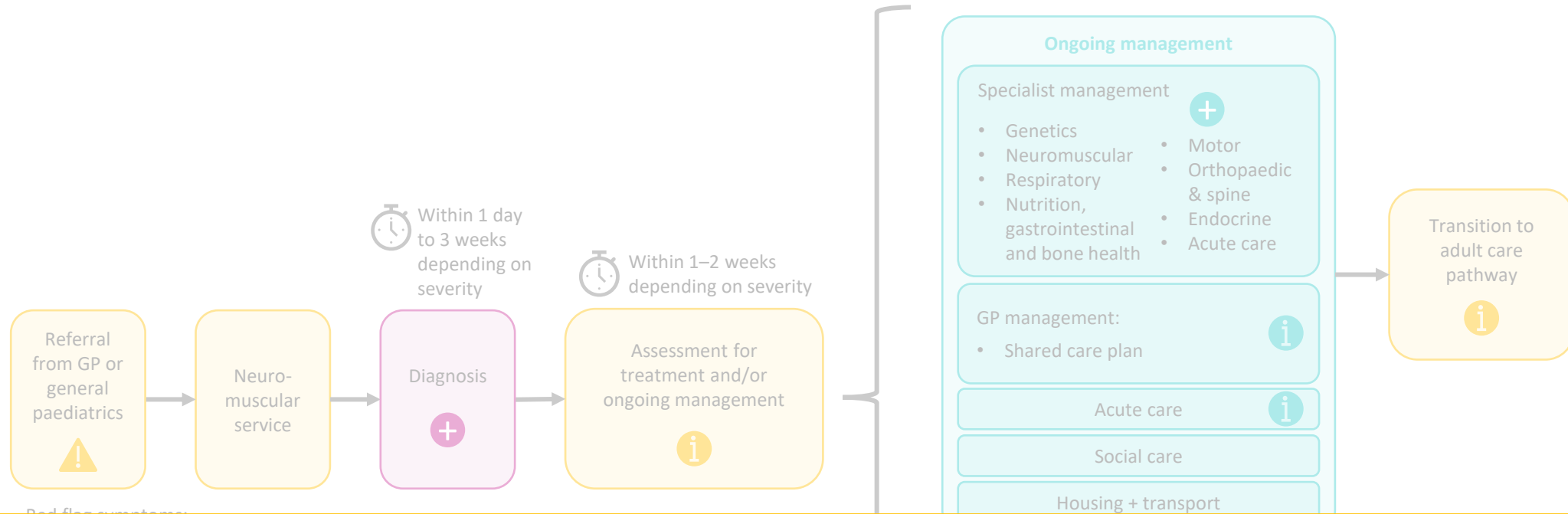
### Psychological support and genetic counselling

What to expect from treatment and management.

Parents' concerns and managing their expectations:

- Will my child be able to walk?
- Will my child's development be comparable to other kids of their age?
- Will my child breathe and eat normally?
- I saw on social media that...





### Children’s palliative care services

Palliative care is an active approach to care that supports physical, emotional and practical needs in order to achieve the best quality of life for both children and families. This includes the provision of short breaks / respite care. Children’s palliative care teams provide care for children with life-limiting conditions in hospitals, hospices and at home. Palliative care teams employ a number of different health and social care professionals, including specialist nurses, support workers and play therapists.





Ongoing management

Specialist management



## 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:

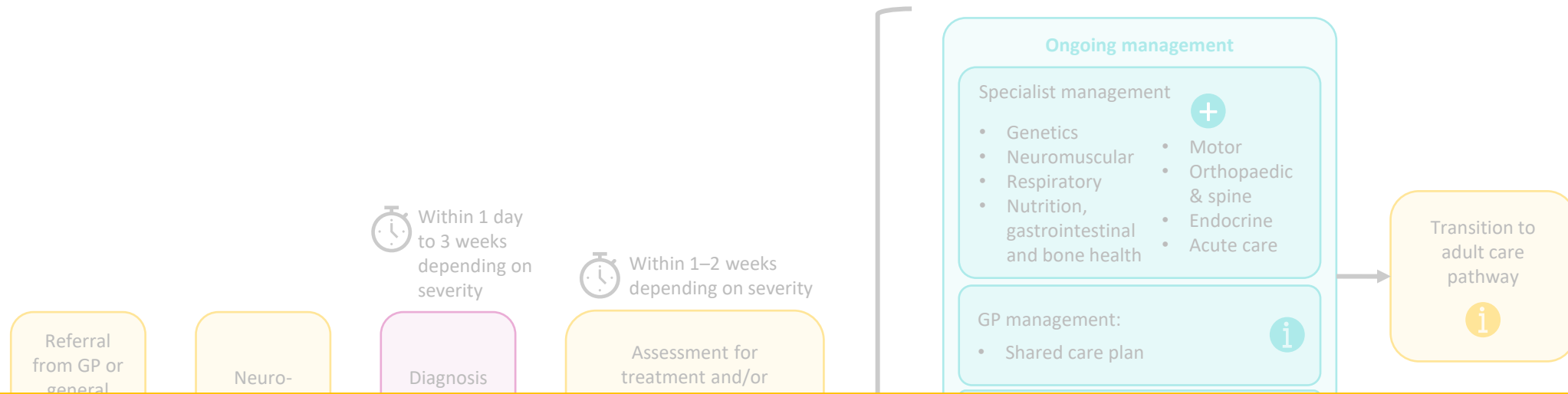
- Vitamin D and nebuliser prescription.
- Importance of vaccinations including the specific types (e.g. RSV vaccine, although a change in national guidance is also needed here).
- Antibiotic prescriptions (GPs need training on the longer period recommend for these patients [10–14 days] and the low threshold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



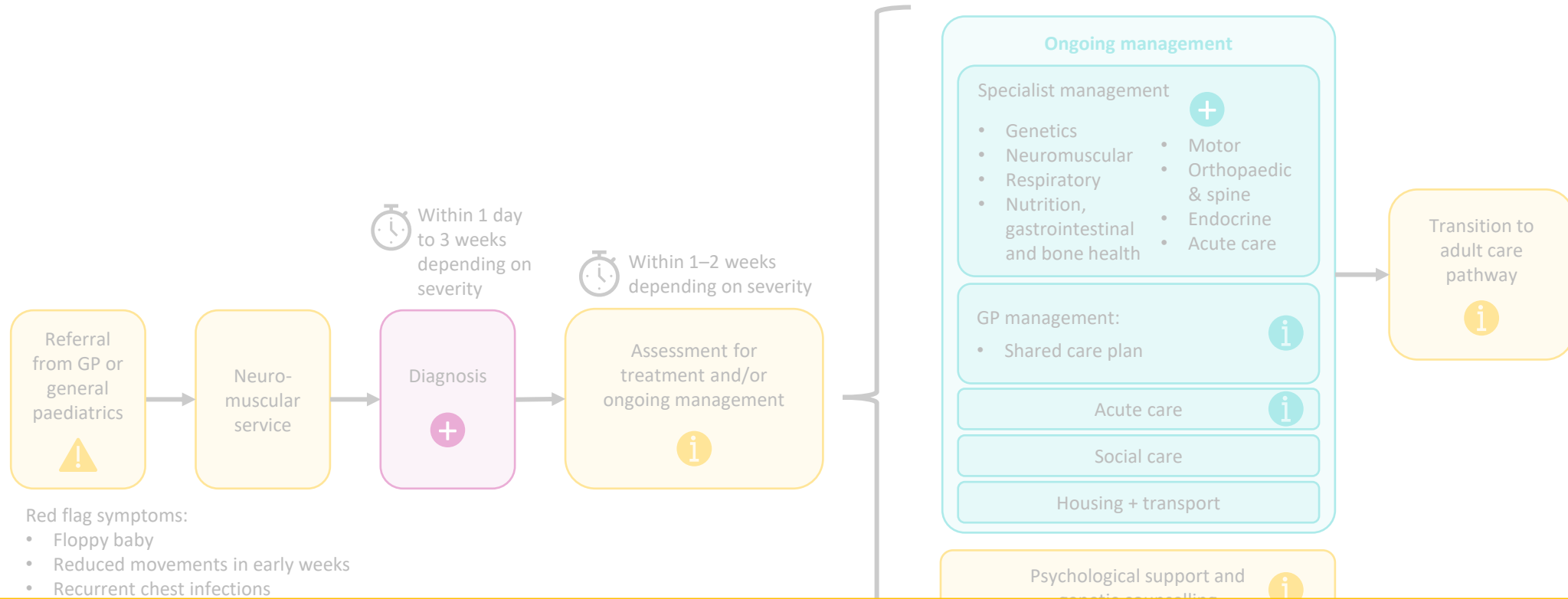
### Transitioning from paediatric to adult care

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.
- Ensure timely introduction of new/alternative equipment

See also: [Care Quality Commission \(2014\) From the pond into the sea: Children’s transition into adult health services](#)





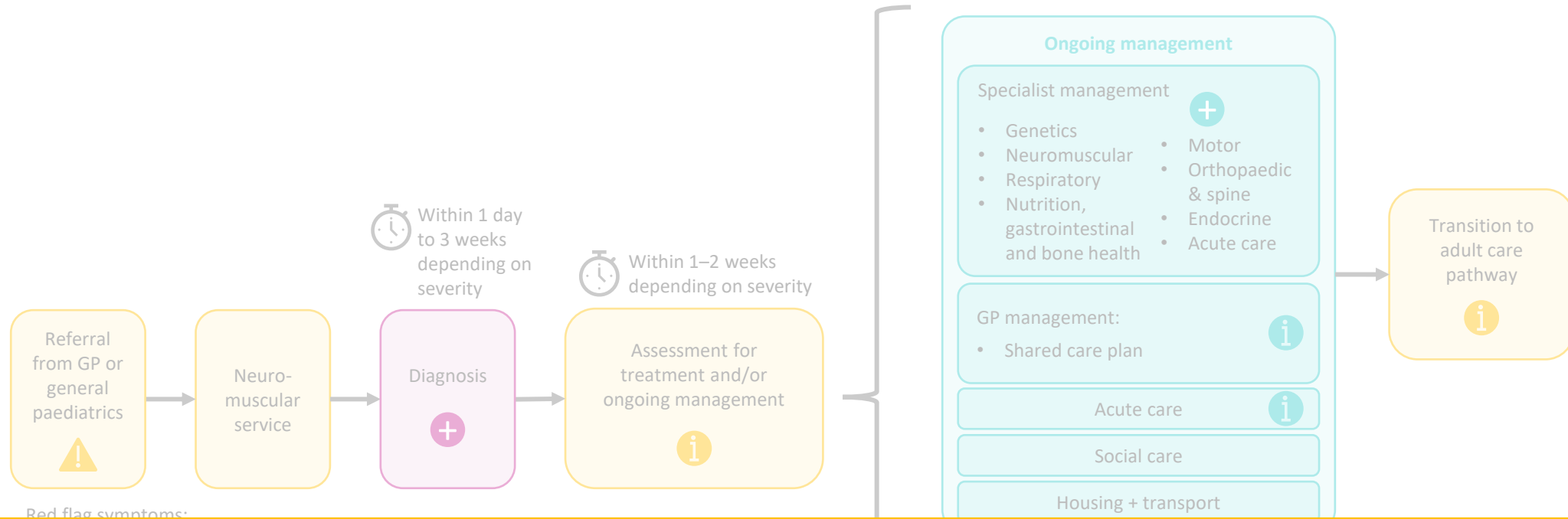
- Red flag symptoms:
- Floppy baby
  - Reduced movements in early weeks
  - Recurrent chest infections



### 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 infants and children 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.



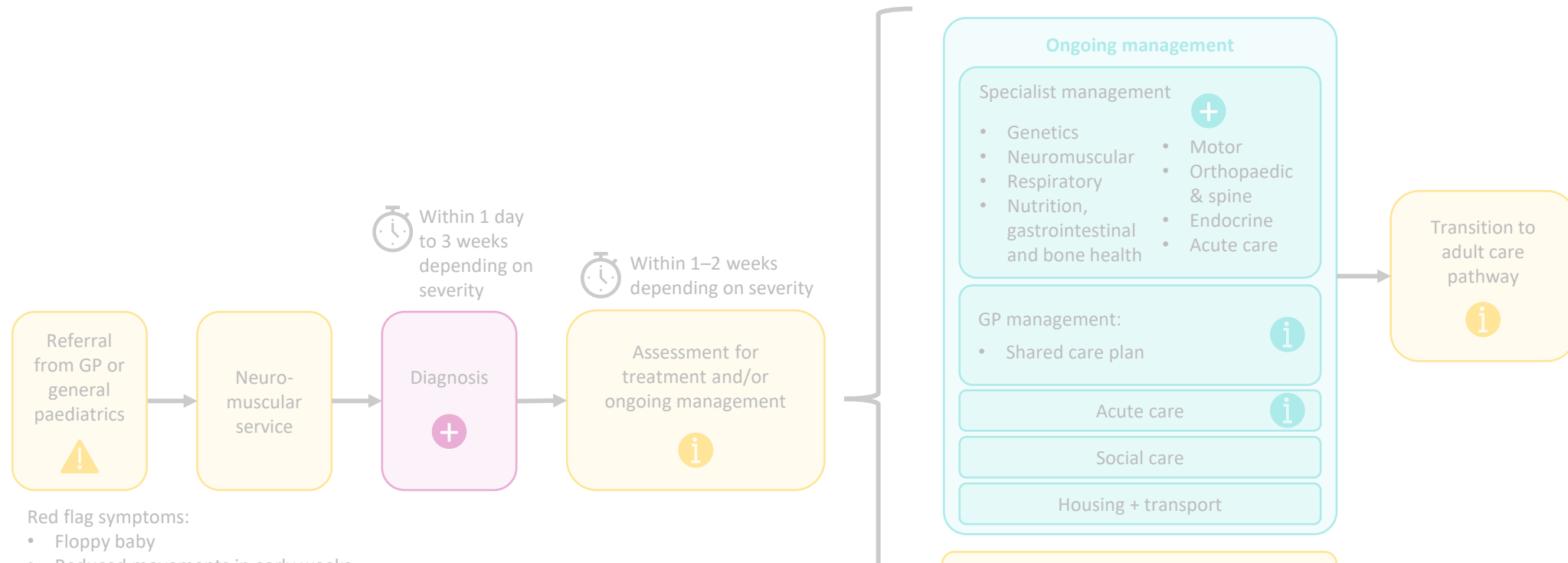


### Audit points

- Timing of referrals:
  - Urgent referral within 1 day to 3 weeks depending on severity.
- Family 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.
  - Infant/child referred for specialist medication (if appropriate) and family received information leaflet about the medication detailing side effects and instructions about monitoring.







### Key performance indicators

- Identified benefit to infant and family.
  - Family has realistic expectations of diagnostic process.
  - Family has understanding of prognosis, available treatment and outcomes.
- Benefit to health professionals and organisation.
  - 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.





## 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](#)

