



IDENTIFICATION, EVALUATION, AND MANAGEMENT OF CHILDREN WITH COMMON NEURO DEVELOPMENTAL DISORDERS -

OTHER THAN AUTISM AND CEREBRAL PALSY

A GUIDELINE FOR THE CLINICIANS AT CHILD
DEVELOPMENT INTERVENTION CENTERS



World Health
Organization



Identification, Evaluation, and Management of Children with Common Neuro Developmental Disorders – other than Autism and Cerebral Palsy

IncluDe

A Guideline for the Clinicians at Child Development Intervention Centers

Child care, Development and Special Needs Unit

Family Health Bureau

Ministry of Health

2022



**World Health
Organization**

Synopsis of IncluDe Logo

The IncluDe logo depicts the concept of ‘embracing diversity’. All children are valuable for our motherland. Children are diverse and their diversity brings different strengths to the development of the country. Hence, as a country we include all the children in any health program. The different shape and coloured letters of the logo symbolizes the diversity. Some may be with disability but they too stand bold. The Inclusive Early Childhood Development program is about all children in Sri Lanka! All are INCLUDED!



Identification, Evaluation and Management of Children with Common Neuro Developmental Disorders: A guideline for the Clinicians at Child Development Intervention Centers

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Message from the Director General of Health Services

Brain development begins during pre-natal stage and extends up to 15 years of age. However, majority of brain development occurs during first three years of life. Hence, identification of child neurodevelopmental disorders and initiation of interventions during this period ensures an optimum outcome in children with neurodevelopmental disorders.

Each and every child have right to attain their full potential of development. For the children with neurodevelopmental disorders to achieve their optimum growth and development health sector has to play a significant role. Identifying such children early as possible through screening and directing them to the multi-sectorial team for further management and follow up is a responsibility of health care workers.

This manual provides all the relevant technical information for clinicians at child development intervention centers on recognizing, managing and following-up children with neurodevelopmental disorders other than autism and cerebral palsy. I hope this guide will be beneficial in the process of early identification and intervention of such children. My warm thanks go to all the contributors for their extreme support in construction of this manual.

Dr. Asela Gunawardena,
Director General of Health Services,
Ministry of Health, Sri Lanka

Message from the Director, Maternal and Child Health

Neurodevelopmental disorders are conditions basically related to the activities of the brain and neurological system. There are many neurodevelopmental disorders of childhood such as attention-deficit hyperactivity disorder (ADHD), cerebral palsy, autism spectrum disorder, learning disabilities and vision/ hearing impairments.

Children with neurodevelopmental disorders may face many difficulties of survival due to issues in language/ speech, motor skills, behavior and memory. With the advancing age features of neurodevelopmental disorders may tend to vary or may remain constant. However, these children with neurodevelopmental disorders should be filtered out from normal children in the society and should provide a multi-disciplinary approach to bring out the best from these children.

This manual has been prepared as a comprehensive guide especially for the clinicians who provide basic care for the children diagnosed with neurodevelopmental disorders. This consists of the appropriate guidelines required to provide care for the children with neurodevelopmental disabilities. I hope this will be maximally utilized by health care workers to brighten up the little lives of children with neurodevelopmental disorders.

Dr. Chithramalee de Silva

Director Maternal and Child Health

Family Health Bureau

Message from the WHO Country Representative

The prevalence of neurodevelopmental disorders are rising around the world. This situation is more or less similar in Sri Lanka. If these children are not addressed on time there is high possibility of these children to develop co-morbidities like conductive disorders, anxiety, mental disorders etc.

Children with neurodevelopmental disorders generally struggle with cognitive, emotional and behavioral problems. This struggle impairs the lives of both child and the family. As most of the time such conditions are permanent and long lasting such a child will have to survive his/her entire life with this struggling. As service providers it is our responsibility to identify them without a delay and absorb them to a system designed to improve their well-being.

In Sri Lanka, with the initiation of IncluDe program, child development intervention centers (CDIC) were established. From these centers a multi-disciplinary approach will be provided to those who are in need. This handbook will provide guidance to the clinicians of CDIC for managing childhood neurodevelopmental disorders except for autism and cerebral palsy which have been elaborated separately.

This handbook is a consequence of numerous contributors' determination and at the end I would like to appreciate all of their efforts.

Dr. Alaka Singh

WHO Country Representative for Sri Lanka

List of Abbreviations

B/L CP	-	Bilateral Cerebral Palsy
CDC	-	Child Development Clinic
CDIC	-	Child Development Intervention Clinic
CDIC-Reg	-	Regional Child Development Intervention Clinic
CHDR	-	Child Health and Development Record
CNS	-	Central Nervous System
CP	-	Cerebral Palsy
CWC	-	Child Welfare Clinic
ECI	-	early childhood interventions
eNIMS	-	Electronic Neonatal Information Management System
ENT	-	Ear, Nose, Throat
ET	-	Educational Therapist
FHB	-	Family Health Bureau
GMA	-	General Movement Assessment
GMFCS	-	Gross Motor Function Classification System
GP	-	General Practitioner
HIV	-	Human Immunodeficiency Virus
HINE	-	Hammersmith Infant Neurological Examination
IIP	-	Individualized intervention plans
IncluDe	-	Inclusive Early Childhood Development Program
LMIC	-	Low Middle-Income Countries
MCH review	-	Maternal and Child Health Review
MDT	-	Multidisciplinary team
MOH	-	Medical Officer of Health
MOH CDC	-	Medical Officer of Health Child Development Clinic
NICU	-	Neonatal Intensive Care Unit
NO	-	Nitric Oxide
OT	-	Occupational Therapists
PBU	-	Premature Baby Unit
PHM	-	Public Health Midwife
PHNS	-	Public Health Nursing Sister
PT	-	Physio Therapist
RDHS office	-	Regional Director of Health Services office
SCBU	-	Special Care Baby Unit
SD	-	Standard Deviation
SGA	-	Small for Gestational Age
SLT	-	Speech and Language Therapists
SPHM	-	Supervising Public Health Midwife
WBC	-	Well Baby Clinic

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Early childhood is a crucial phase of growth and development because experiences during early childhood can influence outcomes across the entire course of an individual's life. Developmental impairments may result from a heterogeneous group of conditions that begin early in life and present with delay or abnormal pattern of progression in one or more of the main development domains, namely motor skills, speech and language, social emotional skills and cognition.

As the aetiology of developmental impairment can be identified in as many as 40-80% of cases, it's vital for the clinicians to evaluate these children for an etiological diagnosis with relevant subspecialties using an integrative approach. Establishing a diagnosis enables clinicians to define treatment options and conduct surveillance for known complications as well as provide prognosis and condition-specific family support.

Yet a larger majority of children will remain undiagnosed for an aetiology but requires well planned management to bring about the best functional outcomes.

It is the right of every child to be able to achieve their full developmental potential. Health sector, among other stakeholders in the society, has a pivotal role to play in providing necessary care and services for children with developmental impairments to reach their maximum potential in growth and development.

Thus, early identification and intervention of children with developmental impairment is critical for better outcome due to the narrow window of *neuronal plasticity*. It is the responsibility of the paediatric professionals to identify such children and absorb them into the healthcare system for further management and follow-up in a multi-disciplinary setting.

The objective of this guideline is to provide a framework for physicians to better manage children with various developmental delays and disabilities. This guideline gives initial introduction and definition of commonly used terms in developmental paediatrics. There is guidance on communication with families and general advice on parenting. This includes guidelines for recognition, assessment, management, referral and follow up care of children with specific developmental impairments (genetic syndromes, speech and language delay, eating, drinking and swallowing impairment, hypotonia, developmental regression, acquired brain injury, spinal dysraphism, visual impairment and under-stimulation). Guidelines for management of Cerebral Palsy and Autism Spectrum Disorder are formulated separately, hence will not be discussed in this guideline.

1.1 Epidemiology

It is estimated that about 10 -15% of children under 5 years of age are having isolated developmental delay while the prevalence of global developmental delay is 1-3%.

Children in the community with developmental delay or disability could be detected in following situations.

- Developmental screening and surveillance of children with identified risk factors (e.g. prematurity).
- During routine child health surveillance or developmental screening
- Parents and caregivers with concerns regarding delayed milestones, challenging behaviours etc, seeking medical advice.
- Professionals in daycare centers or preschool setting may recognize deviant patterns of development and direct parents for medical referral
- Recognition of development impairment opportunistically at health contacts at primary or secondary care levels.

Possible causes of developmental impairments are summarized in table 1- Annexure 1

1.2 Definitions

Early childhood: the period from prenatal development to eight years of age.

Child development: the sequence of physical, cognitive, communication and emotional changes that occur in a child from birth to adulthood.

Child development is usually described under 5 main domains:

1. Gross motor
2. Fine motor
2. Speech and language
3. Socio-emotional
4. Cognition

Developmental delay: Developmental delay refers to children who experience significant variation in the achievement of expected milestones for their actual or adjusted age. The developmental performance more than 1.5 standard deviations below the mean in a standard developmental assessment tool in children under 5 years of age, is defined as a developmental delay.

Global developmental delay: A delay in two or more of main development domains. Significant GDD is when there is a deficit in performance of at least 2 SD below the age appropriate mean on accepted standardised assessment tests.

Early Childhood Intervention: Multidisciplinary services provided to children from birth to 5 years of age to promote child health and well-being, enhance emerging competencies, minimise developmental delays, remediate existing or emerging disabilities, prevent functional deterioration and promote adaptive parenting and overall family function.

Developmental Surveillance: A flexible, continuous process whereby knowledgeable professionals perform skilled observations of children during the provision of health care. The components of developmental surveillance include eliciting and attending to parental concerns, obtaining a relevant developmental history, making accurate and informative observations of children, and sharing opinions and concerns with other relevant professionals.

Developmental Screening: The use of a standardized developmental questionnaire/ brief assessment at regular intervals to search the general population of children to identify children who should receive more intensive diagnosis or assessment delays.

Developmental Evaluation or Assessment: A comprehensive assessment of an at-risk child's developmental level and functioning, using a standardized tool and clinical observations, usually performed by a medical specialty provider or an Early Intervention specialist.

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2.1 Challenges

- Parents suffer a range of emotions and psychological stressors as they did not get the child they wanted
- Parents may over protect or under protect their child
- Parents suffer guilt and attribute those to various religious and other causes.

2.2 How can we help parents to accept their children and be strong?

1. Support them through the stages of grief.

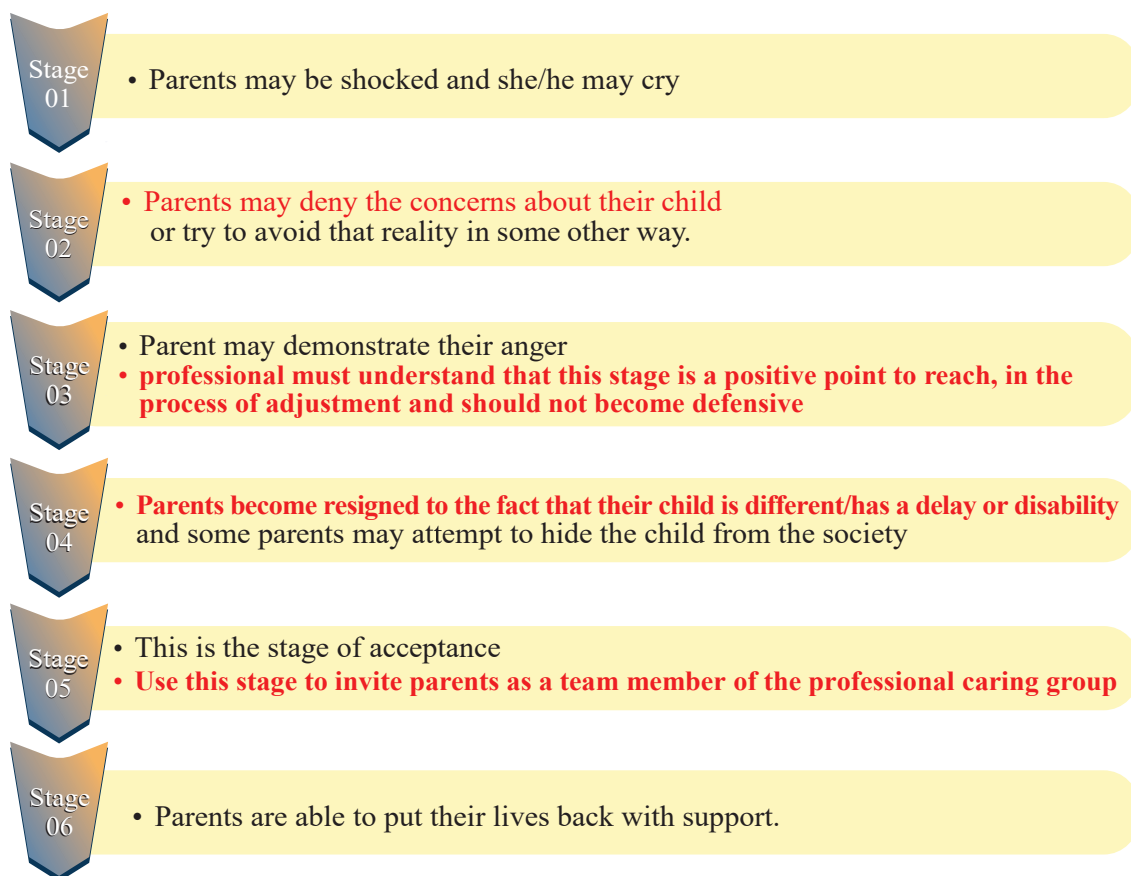


Figure 1: Stages of grief

2.3 Principles of Communicating with parents

Aim is to find a mutually acceptable plan of management and address family concerns.

This is a continuous process in which parents can lead to accept the child as a different rather than lesser

- o Help parents with education about the condition
- o Help through unproductive feelings
- o Connect to resources
- o Help to set realistic expectations
- o Help them to realize child's uniqueness

The diagnosis should be delivered in person, by an available most experienced healthcare professional with sufficient knowledge of the condition.

Healthcare providers should coordinate the message to ensure consistency in the information provided to the family.

Whenever possible, meet with both parents together, if only mother is available, ask to make arrangements to speak with father at a later time.

The family should be informed of the diagnosis in their preferred language. If possible, a professional medical interpreter should be present at the time of disclosure.

Discuss the diagnosis in a private, comfortable setting, free from interruptions. Allow time for questions and make plans for a follow-up conversation.

Parents should be provided with accurate and up-to-date information. Information should be given with a balanced perspective, including both positive aspects and challenges related to clinical condition.

Provide the information in a sensitive and caring, yet confident and straightforward manner, using understandable language that is clear and concise, avoiding medical jargon.

Use neutral language and avoid using value judgments when starting the conversation, such as "I'm sorry" or "Unfortunately, I have bad news".

Use sensitive language and avoid outdated or offensive terminology.

In the newborn setting, the baby should be present, and should be referred to by name. Use person-centric language, emphasizing that this is a baby who has genetic condition.

Allow time for silence and time for tears. Do not feel that you need to talk to "fill the silence". Offer the family time alone.

Assess the emotional reactions of the parents and validate these feelings. Use active listening and empathic responses to support the parents.

Informational resources should be provided, including contact information for local and national support groups, up-to-date printed information or fact sheets, and books.

The opportunity to meet with families who are raising a child with similar condition should be offered if possible.

To reach their full potential, children need the five inter-related and indivisible components of nurturing care: good health, adequate nutrition, safety and security, responsive caregiving and opportunities for learning.

Nurturing children means keeping them safe, healthy and well nourished, paying attention and responding to their needs and interests, encouraging them to explore their environment and interact with caregivers and others.



Terms used for pathways to impact

Promote – Focuses on promoting child development through increasing positive, responsive stimulation between a child and their primary caregiver

Prevent - Reduces exposure to known biological risk factors for adverse developmental outcomes

Protect – Targets population subgroups on the basis of sociodemographic vulnerabilities (e.g. family violence, caregiver mental health)

Figure 2: Components of nurturing care framework

Good health;

- monitoring children's physical and emotional condition;
- giving affectionate and appropriate responses to children's daily needs;
- protecting young children from household and environmental dangers;
- having hygiene practices which minimize infections;
- using promotive and preventive health services;
- seeking care and appropriate treatment for children's illnesses.

Adequate nutrition;

- mother's nutrition and wellbeing during pregnancy,
- exclusive breast feeding till complete six months,
- complementary foods with balance nutrition,
- treat for malnutrition or obesity.

Responsive caregiving;

- protecting children against injury and the negative effects of adversity;
- recognizing and responding to illness;
- enriched learning,
- building trust and social relationships.

Early learning;

- Learning ensures human's successful adaptation to changing circumstances.
- Need to provide opportunity to learn in every possible time.

Security and safety;

- Young children can not protect themselves and are vulnerable to unanticipated danger, physical pain and emotional stress,
- Make sure that defenceless young children feel safe and secure.

3.1 Important points to be remembered on parenting

- Learn what the child's strengths and weaknesses are and how they learn best, what is stressful to the child and what makes him/her happy, and what causes problem behaviours and what prevents them.
- Learn how the child communicates and responds (using words, gestures, non-verbal expression, and behaviours).
- Help the child develop by engaging with her/him in everyday activities and play.
- Children learn best during activities that are fun and positive.
- Involve them in everyday life, starting with simple tasks, one at a time. Break complex activities down into simple steps so that the child can learn and be rewarded one step at a time.
- Make predictable daily routines by scheduling regular times for eating, playing, learning, and sleeping.
- Keep their environment stimulating: avoid leaving the child alone for hours without someone to talk to and limit time spent watching TV and playing electronic games.
- Keep them in the school setting for as long as possible, attending mainstream schools even if only part-time.
- Use balanced discipline. When the child/adolescent does something good, offer a reward. Distract the child/adolescent from things they should not do.
- **“DO NOT” use threats or physical punishment, and never physically abuse the child. Physical punishment can harm the child-carer relationship; it does not work as well as other methods and can make behaviour problems worse.**

Promote and protect the human rights of the person and family and be vigilant about maintaining human rights and dignity.

- Educate carers to avoid institutionalization.
- Promote access to health information and services.
- Promote access to schooling and other forms of education.
- Promote access to occupations.
- Promote participation in family and community life.

Remember.....

- Your child is unique. never compare your child with other children
- Never put down your child in any circumstance, children always need to be empowered
- Give responsibilities, encourage good deeds and discourage or ignore bad deeds.
- Behaviour problems can be an iceberg of an underlying pathology
- When there is a new younger child, elder one is having fear of sharing the love they get from parents, so they can develop attention-seeking behavior or other behavioral issues. During the pregnancy itself tell them that he/she is going to have a sister/brother and tell him that he has a role as the elder sister/brother. Never let him/her feel that he/she has lost his/her attention, love and caring. Get involving him for their new family member's works. Value his ideas and ask his suggestions also.

3.2 What is an emotional outburst ?

Emotional outbursts/meltdown come in all shapes and sizes.

Children can involve expressions of extreme distress, anxiety, frustration or anger that can involve falling to the floor, shouting, crying, kicking, etc.

This is more common between one to three years of age because they have fewer words to express their big emotions. Older children can have emotional outburst too. This can be because they haven't yet learned more appropriate ways to express or manage feelings.



3.2.1 How to manage emotional outbursts/ tantrum?

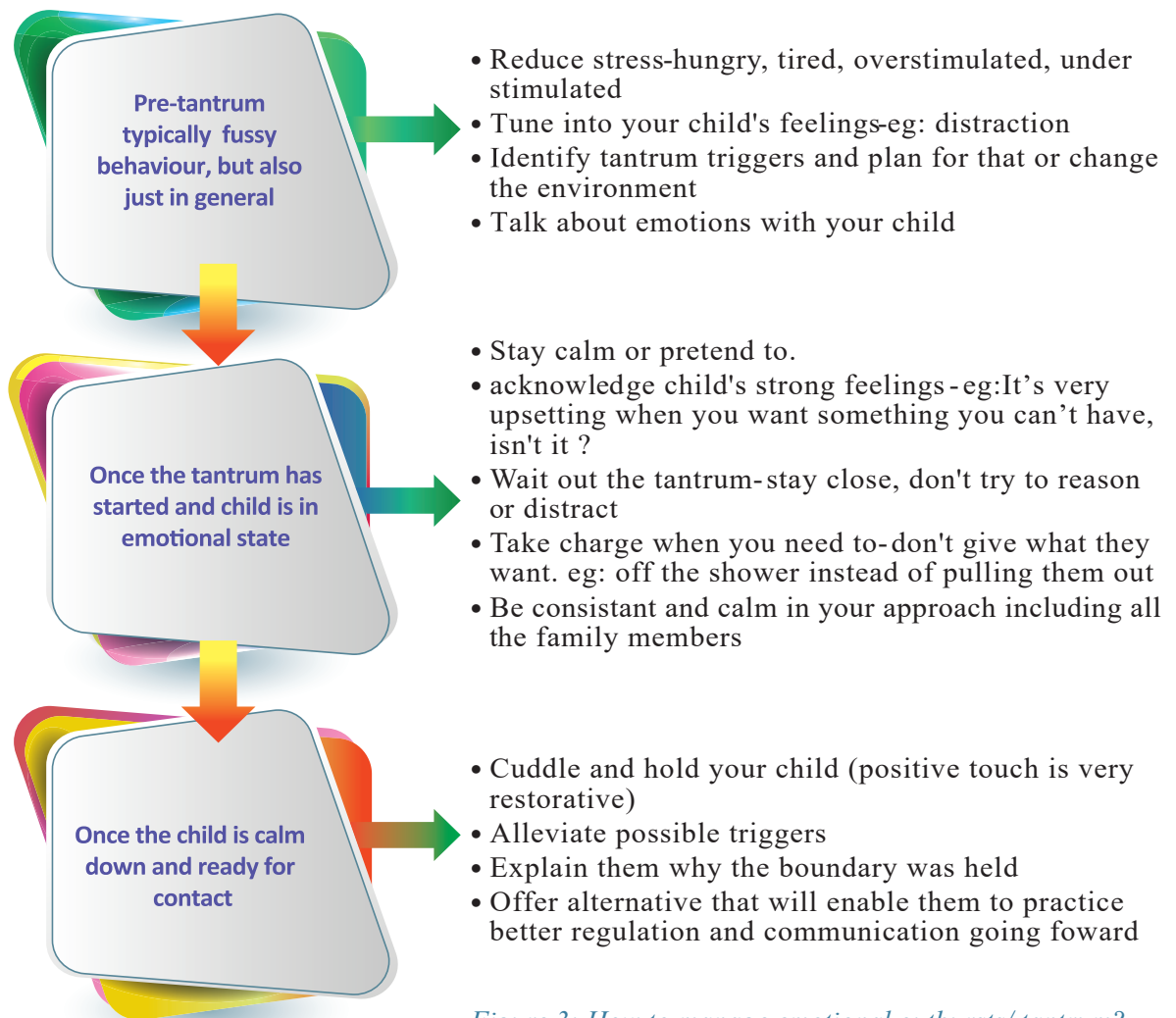


Figure 3: How to manage emotional outbursts/ tantrum?

Reference

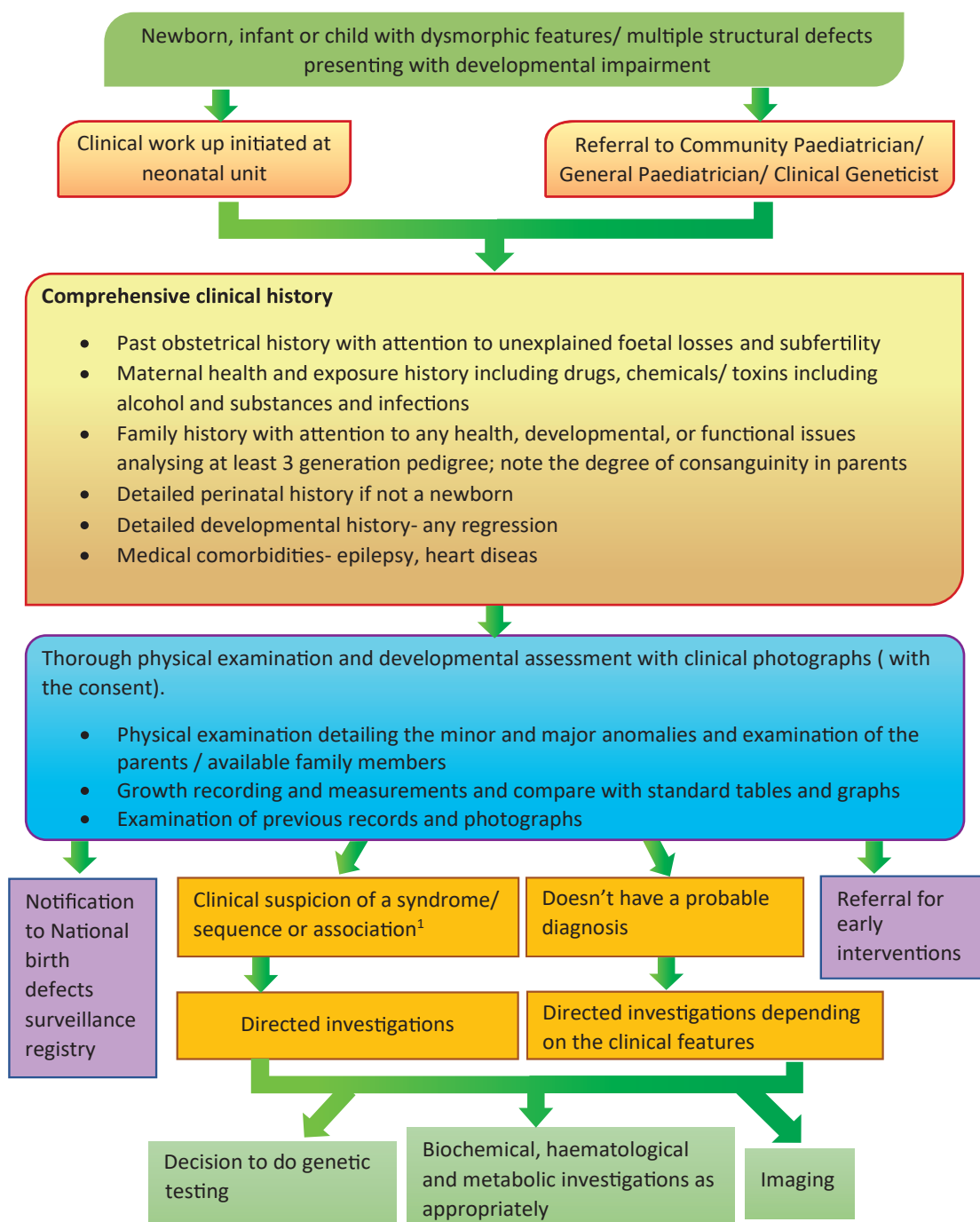
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This guideline discusses several specific neurodevelopmental conditions, focusing on identification, evaluation, management and follow up. Cerebral palsy and autism spectrum disorder are the commonest neurodevelopmental conditions presented in the clinical practice, thus published as separate national guidelines. In the following chapters developmental impairment with genetic aetiology, communication and language impairment, motor and neurological conditions are discussed.

1. Children presenting with neurodevelopmental impairment of a possible genetic aetiology- explains approach, evaluation and available genetic testing in Sri Lanka.
2. Trisomy 21/ Down syndrome- DS is the commonest genetic condition in paediatric age group. This chapter provides guideline for evaluation, follow up and family support for child with DS.
3. Child presenting with language and communication delay- This chapter discusses the pathways of assessing a child presenting with these concerns.
4. Child presenting with hypotonia- Discusses the evaluation and management of a child with hypotonia
5. Child presenting with developmental regression- Discusses evaluation of child presenting with developmental regression and Rett syndrome
6. Management of acquired brain injury
7. Managing and follow up of child with spinal dysraphism
8. Evaluation and managing visual impairment in children- This section gives a guidance on aetiological evaluation and discusses in detail about cerebral visual impairment with the principles of its' management
9. Evaluation of a vulnerable child who could be neglected, under-stimulated or deprived

4.1 Children presenting with neurodevelopmental impairment of a possible genetic aetiology

There are many genetic aetiologies that cause impairment in cognitive/ intellectual, language or motor development. Down syndrome is the most common genetic cause of intellectual disability. Fragile X syndrome is the most common monogenic disorder associated with intellectual disability and is the most common inherited cause for intellectual disability. There are other chromosomal anomalies and microdeletion syndromes that are associated with intellectual disability. Multiple malformation syndromes and inborn errors of metabolism include within other causes of single gene disorders.



¹Malformation sequence: pattern of multiple defects resulting from a single primary malformation, e.g. talipes and hydrocephalus due to neural tube defect.
 Malformation syndrome: pattern of features, often with a unifying underlying cause, e.g. trisomy 21
 Association: group of anomalies that occur more frequently than would be expected by chance alone but that do not have a predictable pattern or unified etiology, e.g. VACTERL association.

Figure 4: Evaluation and management children presenting with neurodevelopmental impairment of a possible genetic aetiology

4.1.1 Genetic Testing

Pre-test counselling

Parents should be informed of the need and potential outcomes after testing.

It should be offered in a way that families are free to refuse or accept according to their wishes and moral beliefs

The best interests of the child should be the primary consideration and relevant ethical issues should be discussed and resolved.

Appropriate counselling and genetic service involvement

For genetic conditions that will not present until adulthood or carrier screening should be deferred until the child is competent to decide whether they want the information and testing.

Table 1: Different types of genetic testing

Genetic testing depending on the suspected genetic aetiology		
Karyotype	Chromosomal aneuploidy, large structural rearrangements, e.g., trisomy 21	Available at Human Genetics Unit (HGU), Faculty of Medicine, Colombo and private sector
Polymerase chain reaction	Fragile X testing Prader Willi syndrome / Angelman Syndrome	
MLPA (Multiplex Ligation-dependent Probe Amplification)	Microdeletion panel, e.g., William syndrome, Di George syndrome	Available at private sector
Whole Exome Sequencing	Suspected Single gene disorders/undiagnosed conditions	Available at Human Genetics Unit (HGU)
DNA microarray Comparative genomic hybridization (CGH)	Detect copy number variations (CNV) in whole genome-microscopic and sub-microscopic imbalances	Not available in SL

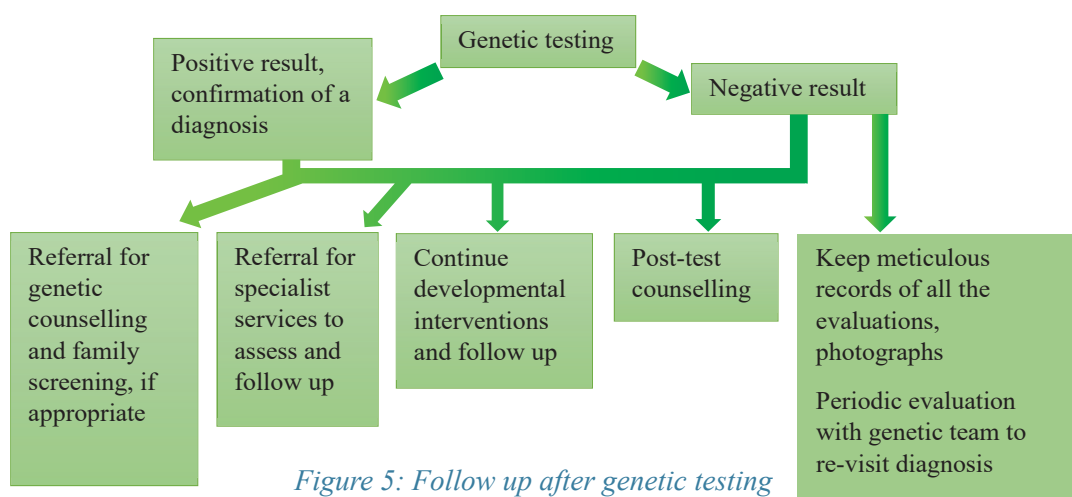


Figure 5: Follow up after genetic testing

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4.2 Down syndrome/ Trisomy 21

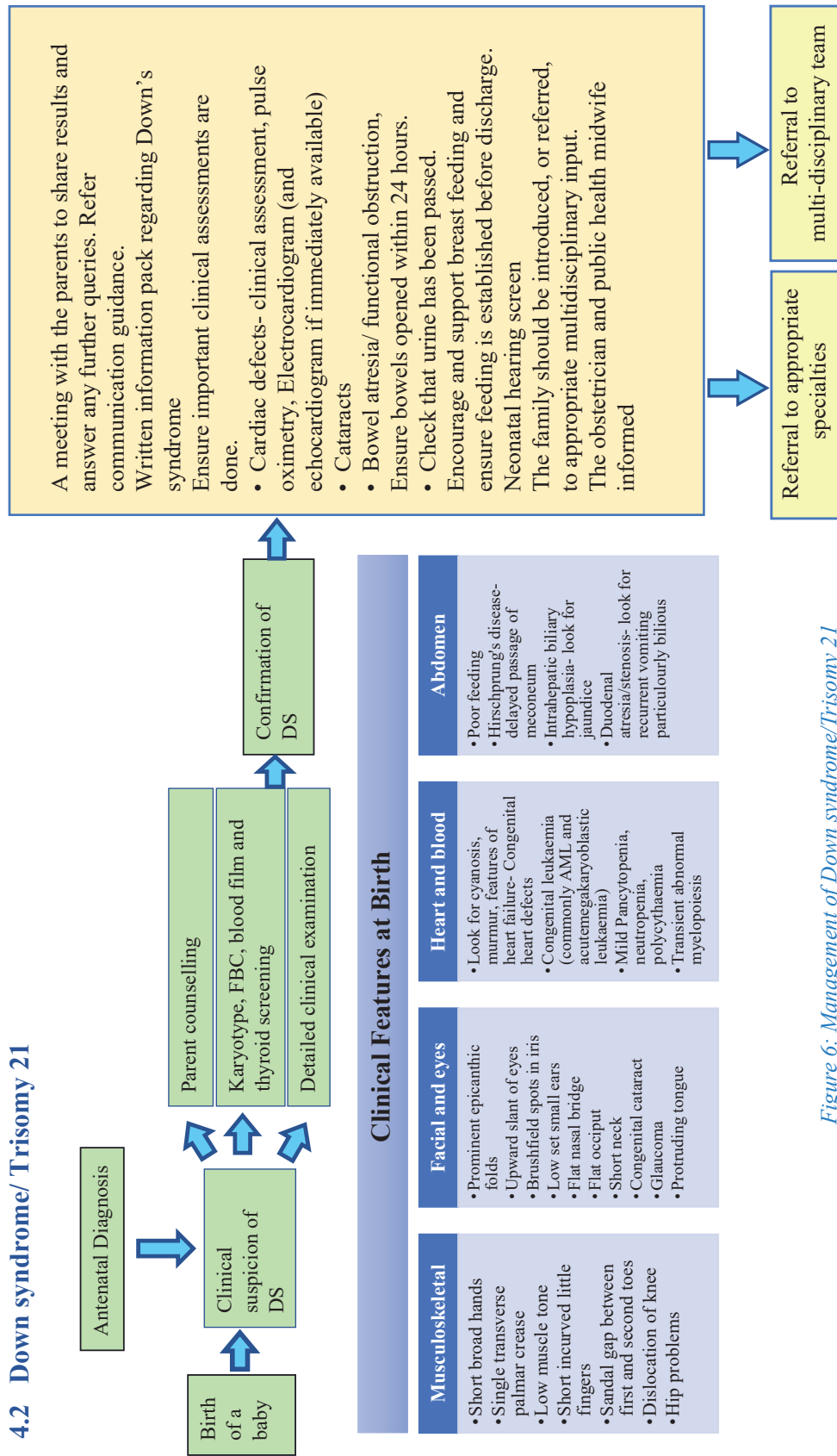


Figure 6: Management of Down syndrome/Trisomy 21



Red Flags within first 6 weeks

- Cyanosis, poor feeding, difficulty in breathing- clinical assessment including SpO₂, ECG – Referral to cardiology
- Jaundice- clinical assessment and consider serum bilirubin, liver functions and USS abdomen
- Delayed passage of meconium, poor feeding, abdominal distention- clinical assessment and consider surgical referral
- Recurrent vomiting, bilious vomiting- clinical assessment and consider USS abdomen and surgical referral
- Poor attachment/ sucking, poor weight gain- assessment of breast feeding
- Poor fixing, absent red reflex- ophthalmology referral
- Poor response to voice- audiology assessment
- Parental concerns and worries- continue parental support, consider mental health support

4.2.1 Recommendations

Recommendation 1: From diagnosis to transition, all children with Down’s syndrome (DS) must be under the care and regular review of a paediatrician with expertise in DS (who may be a community paediatrician, a paediatrician specialising in neuro-disabilities, or a general paediatrician).

Recommendation 2: Children with DS are reviewed by the paediatrician regularly in the first year of life. (A frequency of every 3 months is common practice), and subsequently a minimum of once a year.

Recommendation 3: Specialist services to support the lead paediatrician must be commissioned for all children with DS, namely: Speech & Language Therapy, Paediatric cardiology (echocardiography and clinical review) in the neonatal period, Paediatric ophthalmology, Paediatric audiology. In addition, other professionals must be commissioned to perform assessment and management of complications of DS, if found necessary after review by the DS specialist.

Recommendation 4: A health professional with sufficient knowledge of Down syndrome should communicate at the time of diagnosis, to answer immediate questions, should discuss ideally with both parents. Written information pack regarding Down’s syndrome must be made available to parents immediately on disclosure of diagnosis. The obstetrician and public health midwife should also be informed as soon as possible.

Recommendation 5: The parents and baby should be introduced, or referred, to an appropriate local lead professional (who may be a community paediatrician, a paediatrician specialising in neurodisabilities, or a general paediatrician).for Down syndrome as soon as possible to ensure appropriate multidisciplinary input. Further, refer to caregiver skills training where available.

4.2.2 Follow up

Birth- 6 weeks	6 weeks- 1 year
Early interventions through multi-disciplinary team should be initiated as soon as possible after birth. Regular developmental monitoring and stimulation should be done. Physical therapy for hypotonia and related motor impairment, occupational therapy to support adaptive and fine motor skills, speech and language therapy to support feeding, communication and language development.	
Emotional, social and financial support to family.	
Clinical evaluation for thyroid disorder, TSH and fT4 ¹	Clinical evaluation for thyroid disorder, TSH and fT4 at 6 and 12 months ¹
Clinical cardiac evaluation, pulse oximetry, ECG, 2D echo within 6 weeks	
Clinical eye assessment, red reflex, referral to ophthalmologist	Annual ophthalmological review since 1y
Newborn hearing assessment	Audiological assessment around 6-10 months, Assess and promptly treat middle-ear disease
Growth monitoring using DS specific chart at every well baby visit	Growth monitoring using DS specific chart at every well baby visit
FBC and blood film to assess abnormalities and refer appropriately ²	FBC, ferritin and blood film to assess anaemia at 1 year ³
	Review the signs and symptoms of myelopathy. If present, cervical X ray should be done with appropriate referral. ⁴
Stridor, wheezing, or noisy breathing	
¹Subclinical hypothyroidism (isolated elevation of TSH with normal thyroid hormone levels) is the commonest thyroid hormonal abnormality seen in DS population. Congenital hypothyroidism can be missed if only the fT4 is obtained. Management of children with abnormal TFT should be discussed with a paediatric endocrinologist. Mildly raised TSH (not greater than 10mu/l) or the presence of antibodies with normal T4 and no clinical evidence of hypothyroidism does not usually warrant treatment. Such children should therefore be tested more frequently than those with normal test results. ² Transient leukaemia of Down syndrome (TL-DS)/ transient myeloproliferative disorder (TMD)/ transient abnormal myelopoiesis (TAM) is relatively common (10%). TMD usually regresses spontaneously within the first 3 months of life, but there is an increased risk of later onset of leukaemia for these patients (10%–30%). Parents of infants with TMD should be counselled regarding the risk of leukaemia and made aware of the signs (easy bruising, petechiae, lethargy, or change in feeding patterns). 1-2% will develop acute leukaemia. The most common type seen is myeloid leukaemia of Down syndrome, Polycythaemia is common in infants with Down syndrome (18%–64%) and may require careful management. Definition of TL-DS: Presence of a GATA1 mutation + peripheral blood blast percentage >10% and/or clinical features suggestive of TL-DS (organomegaly, cholestasis and hepatopathy, skin rash, pericardial and pleural effusions) in a child with DS.	

1-5 years	School age
Continue input from MDT. Assess school readiness. Early DS specific and generic educational interventions should be introduced.	Continue input from MDT. Early DS specific and generic educational interventions should be introduced.
Emotional, social and financial support to family. Refer to caregiver skills training, where available	
Clinical evaluation for thyroid disorder, Annual TSH and fT4. AI hyperthyroidism seen in late childhood	
Annual clinical cardiac evaluation for acquired HD and refer appropriately. Children with heart lesions are at increased risk of infective endocarditis	Annual clinical cardiac evaluation for acquired HD and refer appropriately. From late adolescence onwards there is an increased incidence of asymptomatic mitral valve prolapse (MVP) and of aortic regurgitation (AR).
Annual ophthalmological review or as appropriate	Two yearly ophthalmological review or as appropriate
Audiological assessment annually, Assess and promptly treat middle-ear disease	Two yearly audiology assessment or as appropriate, Assess and promptly treat middle-ear disease
Annual growth monitoring using DS specific chart	
Annual FBC, blood film +/- ferritin to assess anaemia. Consider coeliac screening.	
Review the signs and symptoms of myelopathy. If present, cervical X ray should be done with appropriate referral.*	
Annual clinical assessment for sleep disordered breathing	
Ensure good dental health	
*Discuss with parents the importance of cervical spine-positioning precautions to avoid excessive extension or flexion to protect the cervical spine during any anaesthetic, surgical, or radiographic procedure. Parents should be advised that participation in some sports, including contact sports such as football, rugby and gymnastics places children at increased risk of spinal cord injury. Warning symptoms and signs: Deterioration of gait and/or frequent falls, change in bowel or bladder function, neck pain, reduced neck movements, abnormal head posture, torticollis, increasing fatigability on walking, change in general function, deterioration of manipulative skills or weakness. Any child with these features should undergo plain cervical spine radiography in the neutral position and if having a mobile neck, in flexed and extended positions. If significant radiographic abnormalities are present child should be urgently referred to a paediatric neurosurgeon. Child must be 3 years of age to have adequate vertebral mineralization and epiphyseal development for accurate radiographic evaluation of the cervical spine. Routine radiologic evaluation of the cervical spine in asymptomatic children is not recommended.	

Figure 7: Follow up of a child with Down syndrome

4.3 Children with speech and language delays

Speech and language is one of the unique characteristics of human beings, and an essential developmental domain to achieve the full potential to function as an independent adult. Language is a rule governed behaviour. It is made up of a set of sounds and written symbol system used for communication through talking or writing according to cultures and geographies. Communication is the active process of exchanging information and ideas. Communication involves both understanding and expression. Forms of expressions may include personalized movements, gestures, objectives, vocalizations, verbalizations, signs, pictures, symbols, printed words, and augmentative and alternative communication (AAC) systems.

Speech and language delay is one of the most common presentations of developmental impairments or disorders. Feeding difficulties resulting from the oro-sensory and motor pathway deficits may often accompany manifestations of these impairments. Speech and language pathologies will also be a component of delays in motor, cognitive, social emotional and adaptive skills due to the close interplay required for the development of this unique human characteristic together with other domains of development.

Developmental disorders of speech and language are diagnosed when normal patterns of language acquisition are disturbed from the early stages of development. Expressive language disorder is a specific developmental disorder in which the child's ability to use expressive spoken language is markedly below the appropriate level for its mental age, but in which language comprehension is within normal limits. There may or may not be abnormalities in articulation. Receptive language disorder is a specific developmental disorder in which the child's understanding of language is below the appropriate level for its mental age

4.3.1 Common presentations/ red flags

1. Difficulties in early feeding
2. Delayed smile
3. Delayed/ absence of responses to sounds
4. No/ minimal babbling
5. No/ minimal words
6. Mispronunciations/ difficulties in speech
7. Learning difficulties

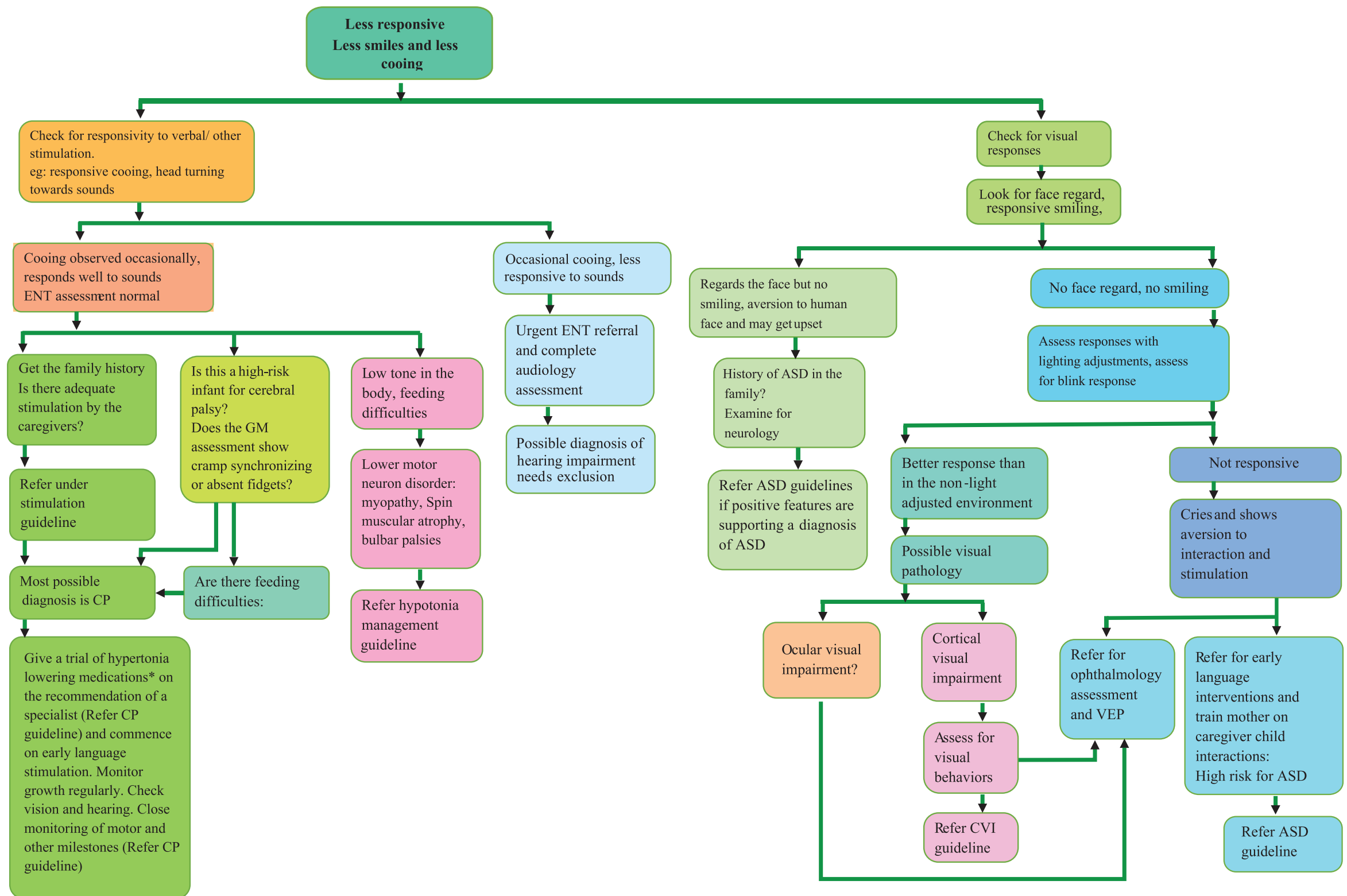
4.3.2 Early concerns in smiling and sound production

Responsive smiling is a universal early communication milestone in human babies established by four months of age. Early concerns regarding smiling could be red flags for two major neuro developmental disorders; ASD and visual impairment. Responsive cooing is also an early communication milestone that will reflect appropriate social and communication development. Therefore, lack of early responsive sound production in terms of cooing and babbling will also point towards ASD and hearing impairment.

All these children need complete and thorough assessments by the ENT, ophthalmology and speech and language clinicians. A neurology opinion when required should always be sought.

4.3.3 A toddler or child presenting with speech delay or speech and language difficulties

This is the commonest presentation by parents. While the entire purpose of the community-based screening programme is to minimise such presentations and for early detection this will continue to be a significant problem always. Families will always notice the difficulties and delays in children first, and it is imperative to build up child development literacy among the public to promote this. Regression of language skills is not discussed in this algorithm as it is addressed separately.



*Trihexyphenidyl
or Syndopa

Figure 8: Early concerns in smiling and sound production

A) Children presenting with speech delay

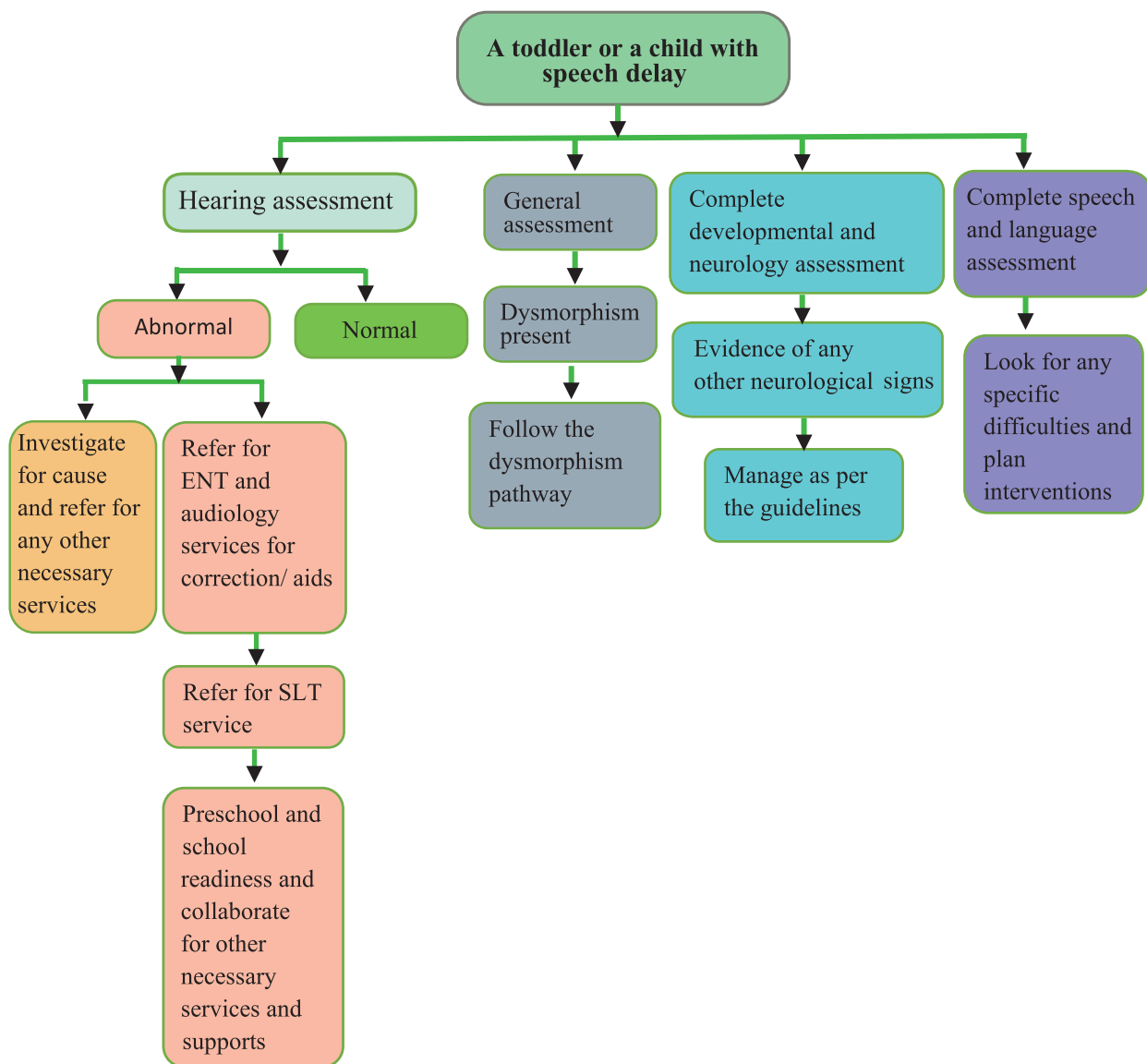


Figure 9: Management of children presenting with speech delay

B) A child presenting with speech difficulties/ unclear speech

Some children may develop early language and communication skills in appropriate sequence but with concerns in quality of the skills. Neurological causes such as brain tumours and neurodegenerative disorders also can result in such manifestations. Also this could be a presenting symptom of other neuro developmental disorders such as cerebral palsy, ASD or learning difficulties which will require long term management by the community team. It is essential to get other specialists involved as required.

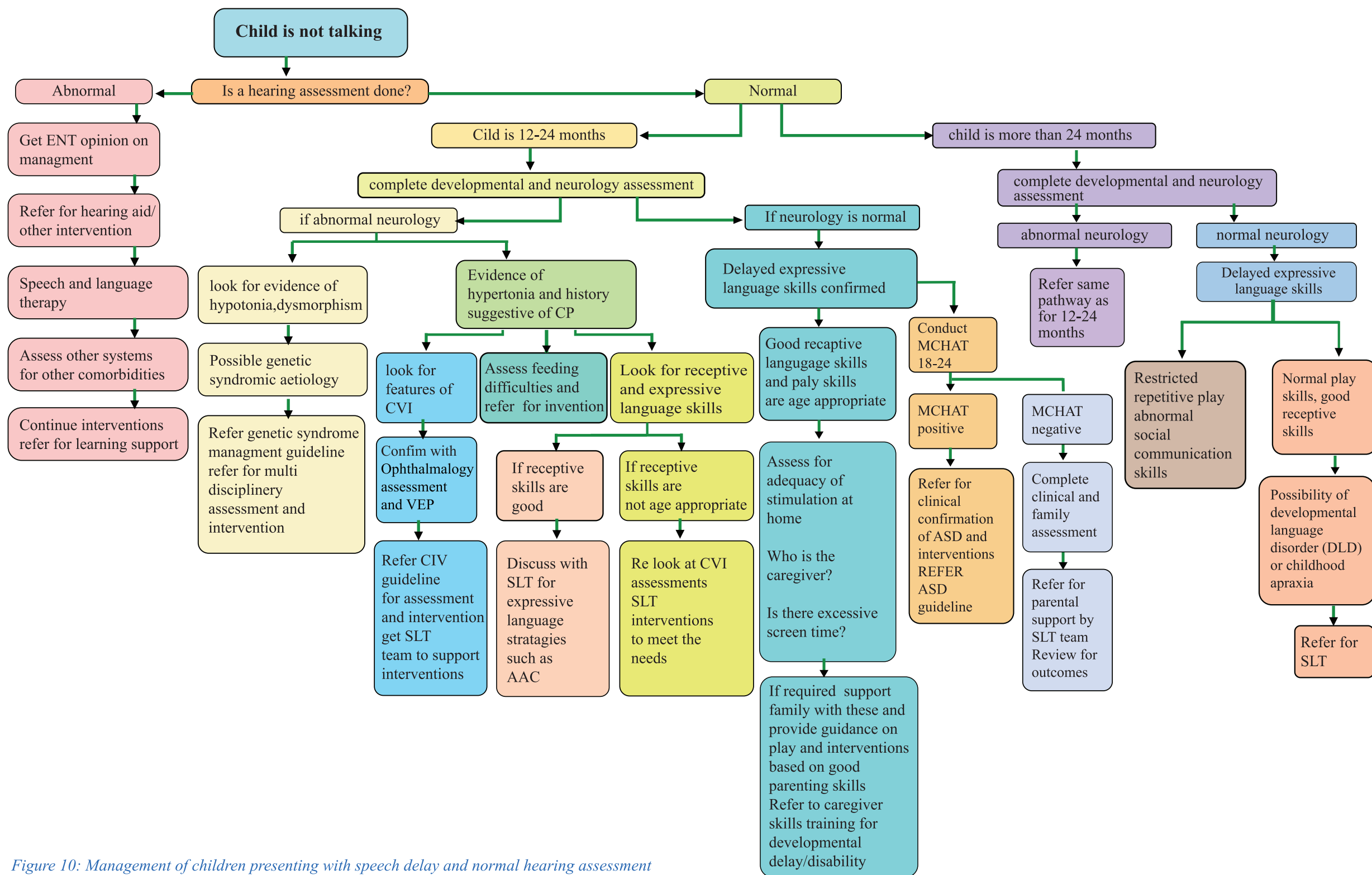


Figure 10: Management of children presenting with speech delay and normal hearing assessment

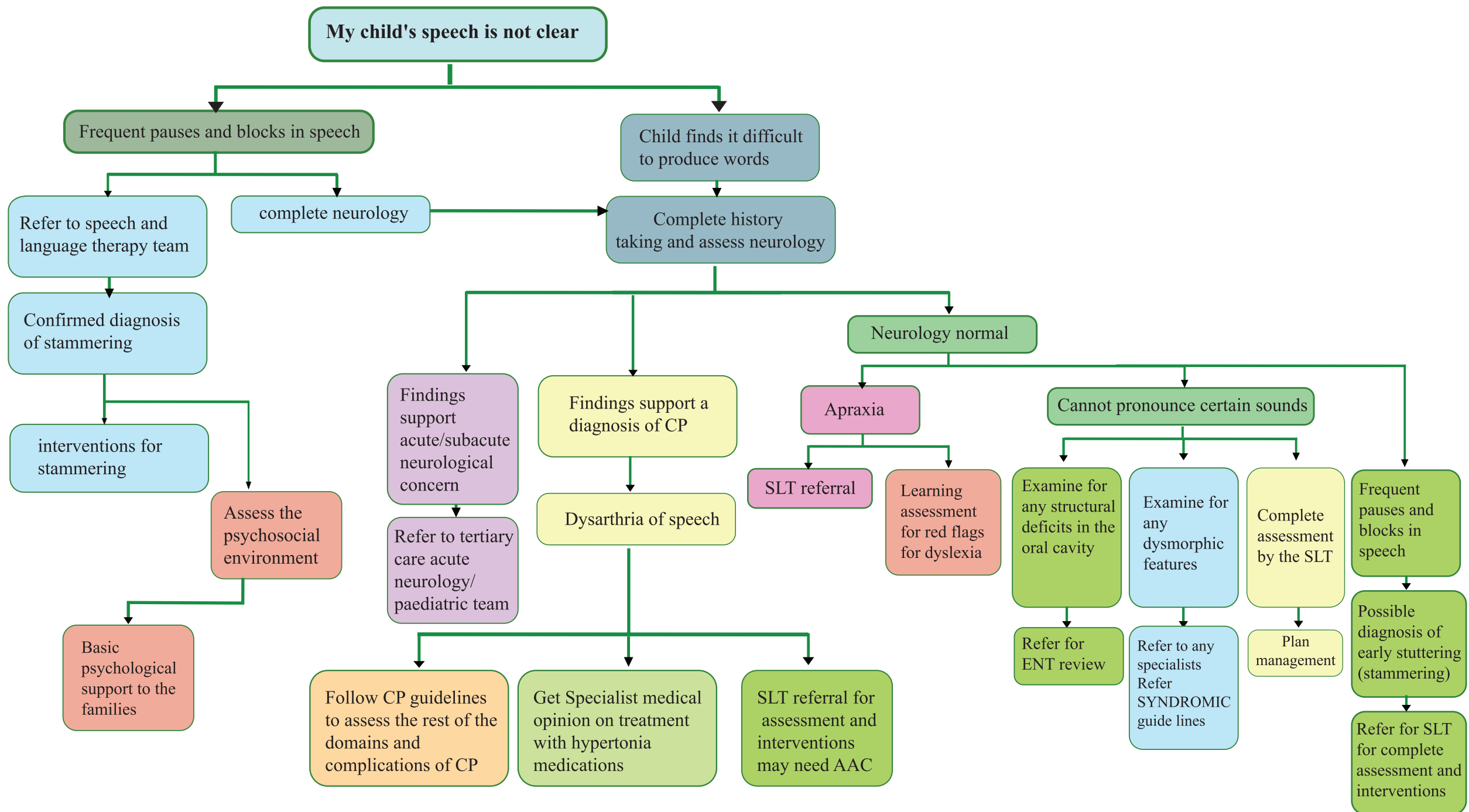


Figure 11: Management of children presenting with speech difficulties/unclear speech

4.4 Early feeding difficulties.

There is a significant association with structural or functional oro- motor difficulties. The golden rule of history and examination should always be accompanied by a thorough oral motor examination in a well- lit environment and should be complimented by the opinion from an ENT specialist if indicated. It is also vital to conduct a detailed observation of the breast-feeding process by a skilled health worker and a complete feeding assessment by a qualified speech and language therapist.

A complete neurological evaluation will help to rule out/ establish a neurological cause for the feeding difficulty and warrants a neurology opinion and further investigations to confirm the pathology. Early feeding difficulties is also a well-recognized manifestation of ASD. Therefore, it is essential that the community teams undertake regular developmental surveillance of all infants who present with feeding difficulties at 6, 12 and 18 months and complete the screening for ASD at 18 and 24 months prior to discharge from care.

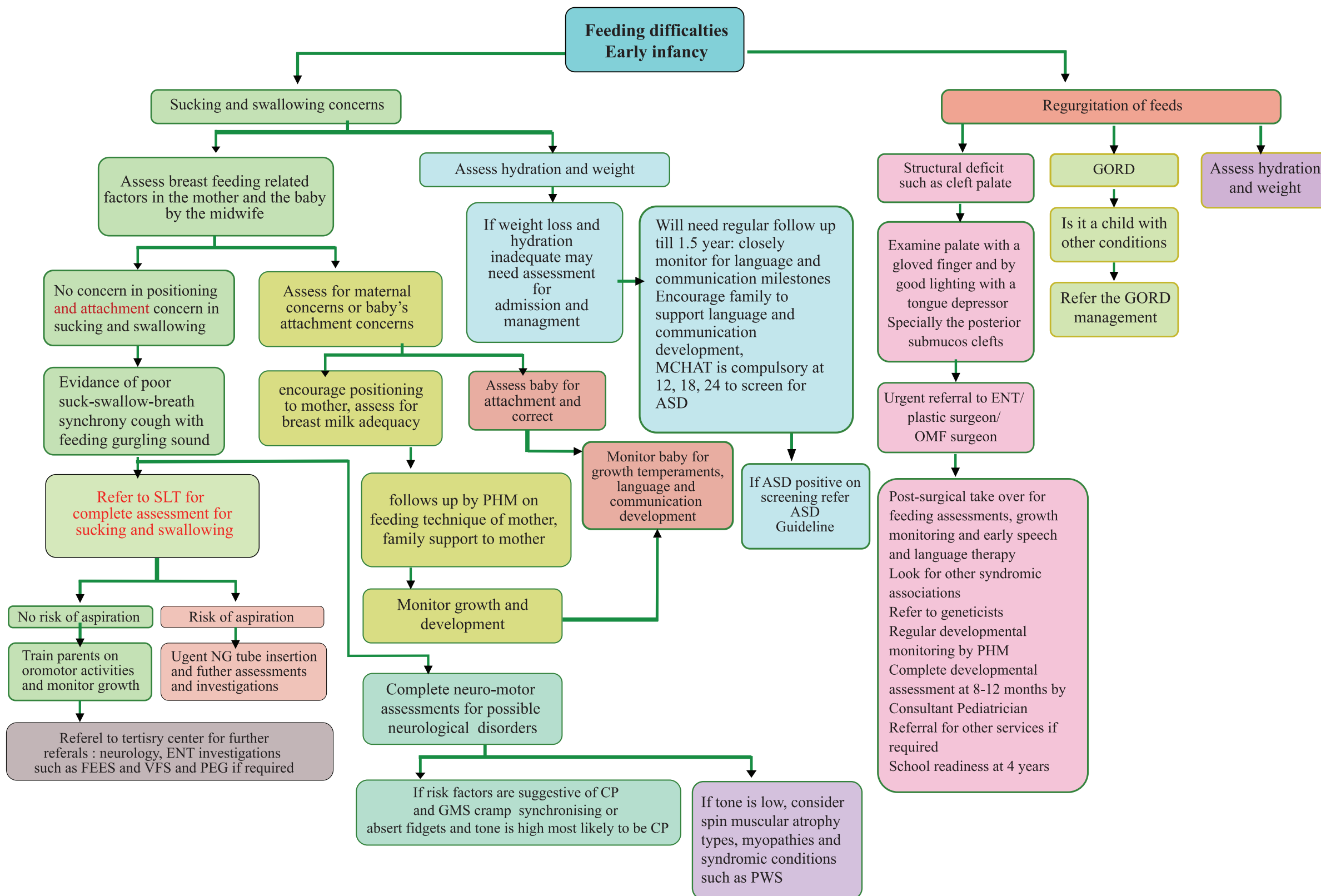


Figure 12: Management of early feeding difficulties

4.5 Evaluation of a floppy child

4.5.1 Differentiating hypotonia and weakness

Definition

Hypotonia is diminished resistance of muscles to passive stretching resulting in:-

- Impaired ability to sustain postural control
- Increased range of movement of joints
- with /without muscle weakness

Weakness - reduction in the maximum power that can be generated

4.5.2 Differential diagnosis of hypotonia

According to the anatomical pathology:-

1. Central Hypotonia (Upper motor neuron type /Strong floppy)
2. Peripheral Hypotonia (Lower motor neuron type /Weak floppy)
3. Combined Central and Peripheral Hypotonia

4.5.3 Causes of hypotonia (Annexure1)

4.5.4 Common presentations



- Weak cry, poor sucking, feeding difficulty
- Development delay - poor head control, delayed sitting or walking
- Feeling limp when lifting
- Having increased flexibility in their joints
- Poor posture
- Tiring easily (have low endurance)
- Recurrent respiratory tract infections
- Seizures

4.5.5 Approach to a child with hypotonia

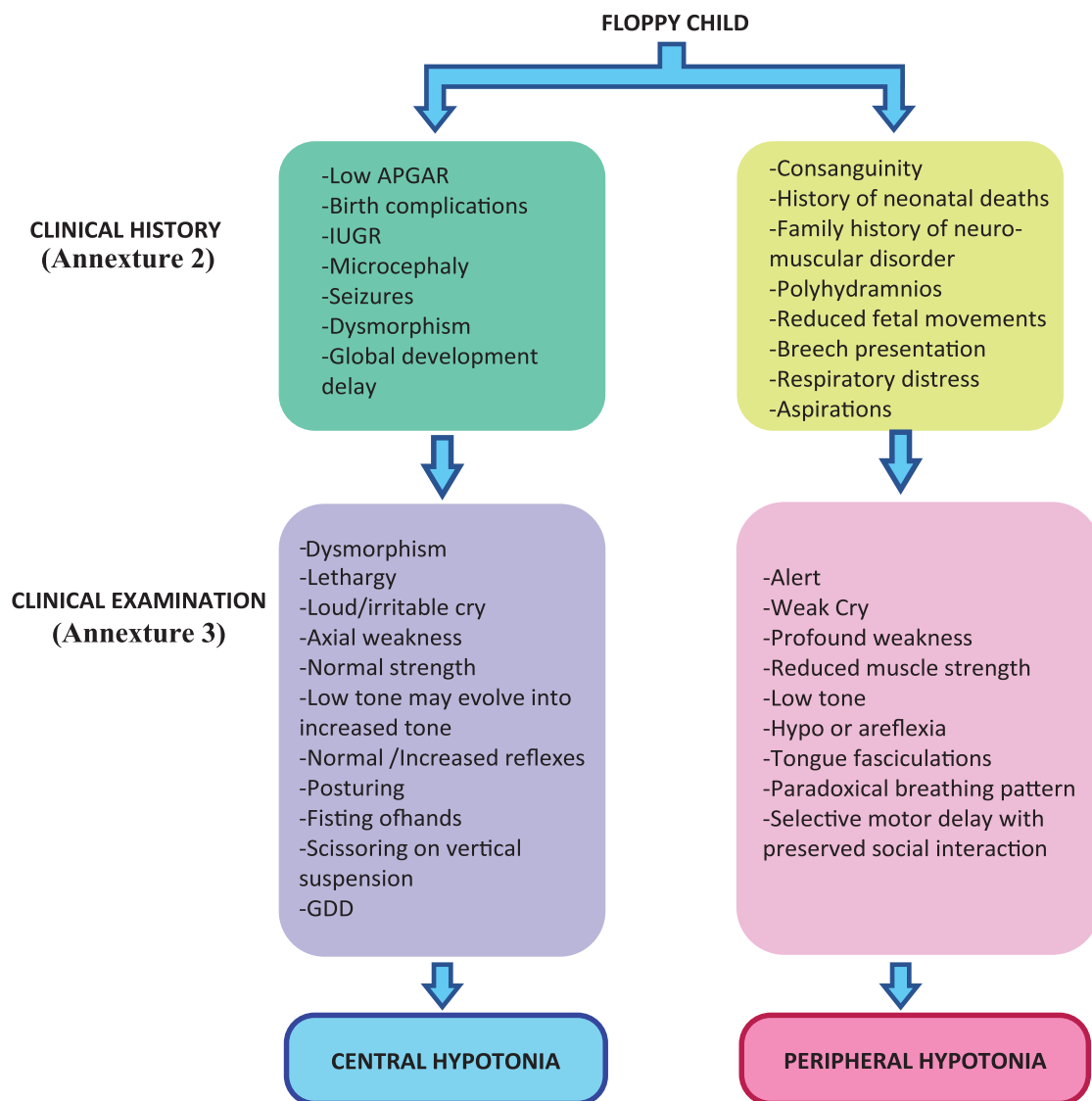


Figure 13: Evaluation of a child with hypotonia

4.5.6 Management

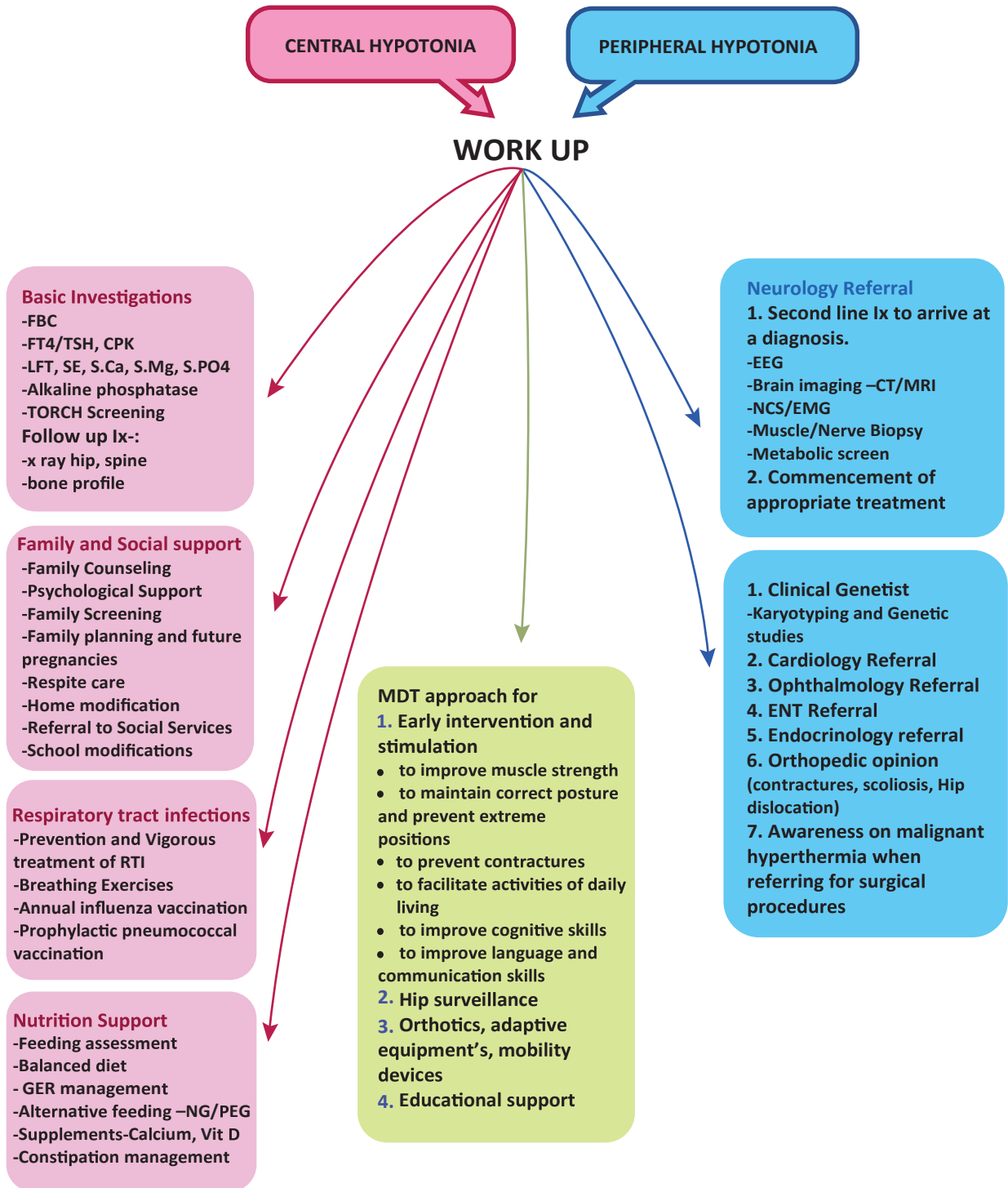


Figure 14: Management of a child with hypotonia

4.6 Neuro - Regressive Disorders

Developmental regression is an important red flag in any child's developmental performance and should be recognized promptly. It is the loss of previously acquired skills and can affect any sphere of childhood development. Presentation and underlying causes are heterogeneous in nature, in turn presenting many clinical challenges.

Developmental regression occurs when children lose certain developmental skills that they had previously successfully acquired. Essentially, they move backwards in their development. In contrast, developmental delay is when children reach milestones later than expected, or experience plateaus in their skills. Developmental regression and developmental delay can occur in one or multiple skills domains. It is important to recognise the key feature of symptom progression before a child is labelled as showing true regression of milestones. Children with significant underlying neurodegenerative pathology will invariably show progressive, ongoing loss of previously acquired skills as time goes by, with no recovery or catch-up.

The causes of regression can be categorised based on clinical findings, so that investigation and management options can be tailored appropriately. It is extremely important to try and make a definitive diagnosis if possible. This allows appropriate initiation of treatment for potentially reversible causes and for specific therapies, which may alter disease progression and improve quality of life in some cases.

*British Paediatric Surveillance Unit/ progressive intellectual and neurological
deterioration study case definition*

Any child (under 16 years of age at onset of symptoms) who fulfils all of the following three criteria:

- ▶ Progressive deterioration for >3 months.
- ▶ Loss of already attained intellectual/developmental abilities.
- ▶ Development of abnormal neurological signs.

Excluding: Static intellectual loss, eg, after encephalitis, head injury or near drowning. Including: Children who meet the case definition even if specific neurological diagnoses have been made

4.6.1 Aetiology

Neurometabolic

1. Storage disorders

- Peroxisomal: X linked adrenoleukodystrophy, Zellweger syndrome, Refsum disease.
- Sphingolipidoses: neuronal ceroid lipofuscinosis (NCL), (Batten disease), Niemann-Pick disease, Krabbe disease, GM1 gangliosidosis.
- GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases), metochromatic leukodystrophy, Gaucher disease.
- Mucopolysaccharidosis (MPS): types I, II and III

2. Transport and processing disorders

- Neurodegeneration with brain iron accumulation, Menkes syndrome, Wilson disease, glutaric aciduria type I, homocystinuria, guanidinoacetate methyltransferase deficiency – Canavan disease, biotinidase deficiency.

3. Mitochondrial Disorders

- Leigh syndrome, myoclonic epilepsy with ragged red fibres, progressive infantile poliodystrophy, neuropathy, ataxia and retinitis pigmentosa

Acquired

- HIV encephalopathy
- Creutzfeldt-Jakob disease
- Subacute sclerosing panencephalitis
- Rasmussen encephalitis
- Autoimmune encephalitis

Neurogenic

- Rett syndrome, Alexander disease
Lafora body disease,
Pelizaeus-Merzbacher syndrome
vanishing white matter disease
Cockayne syndrome
Aicardi-Goutieres syndrome
Huntington's disease

Figure 15: Aetiology of Neuro-Regressive Disorders

4.6.2 Specific pointers in history

Table 2: Specific pointers in history of a child with a neuro-regressive disorder

History	Points to be asked
Present history	Age of onset
	Seizures
	Cognitive impairment & deterioration in school performance
	Gait disturbances: spasticity, ataxia, bradykinesia, dystonia
	Personality and behavioural change
	Vomiting, Hypoglycemia , poor feeding, failure to thrive
	Exaggerated startle response
	Loss of hand skills, hand wringing
	Feeding difficulties
Developmental history	It's important to note whether child is having global / regression or predominant domain of development is affected
Family history	Consanguinity
	Early onset deaths
	Previous affected siblings

4.6.3 Physical examination

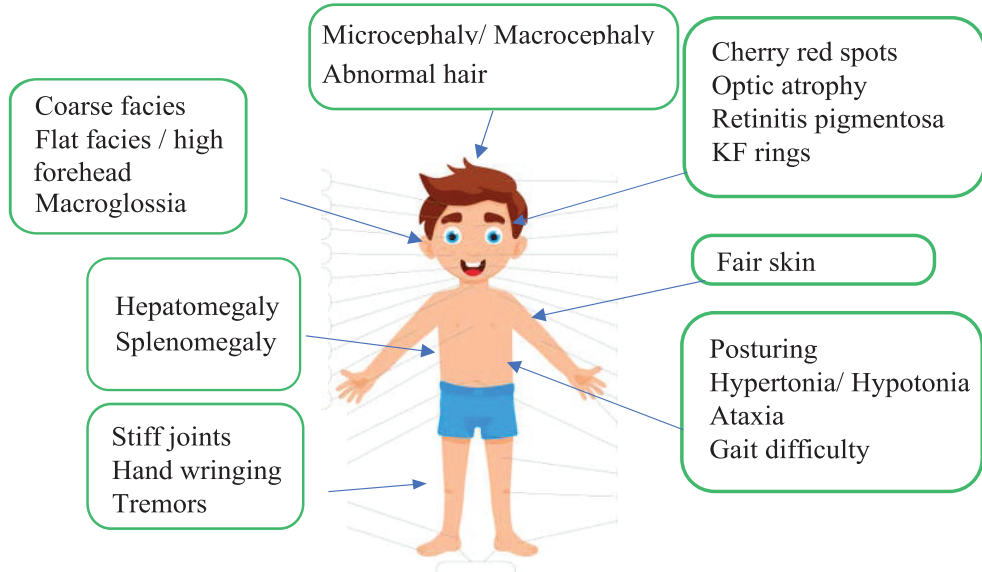


Figure 16: Physical examination of a child with neuro-regressive disorder

4.6.3.1 Clues on examination

- ▶ Head circumference: macrocephaly (white matter disorders), microcephaly (grey matter disorders)
- ▶ Dysmorphic features: mucopolysaccharidosis, Zellweger syndrome
- ▶ Ophthalmology: ptosis (mitochondrial disorders, Menkes syndrome), retinal degeneration (grey matter disease), optic atrophy (white matter disease), cherry-red spots (GM1/2 gangliosidosis, Krabbe leukodystrophy, metachromatic leukodystrophy)
- ▶ Neurology: changes in tone and coordination, features of raised intracranial pressure
- ▶ Skin: port-wine stain (Sturge-Weber), café-au-lait spots and freckling (neurofibromatosis type 1), ash-leaf macules, Shagreen patch and adenoma sebaceum (tuberous sclerosis), abnormal 'kinky' hair (Menkes syndrome), dry skin/hair (biotinidase deficiency)
- ▶ Other systemic clues: visceromegaly (notably hepatosplenomegaly), skeletal signs (eg, dysostoses)

4.6.4 Development regression pathway algorithm

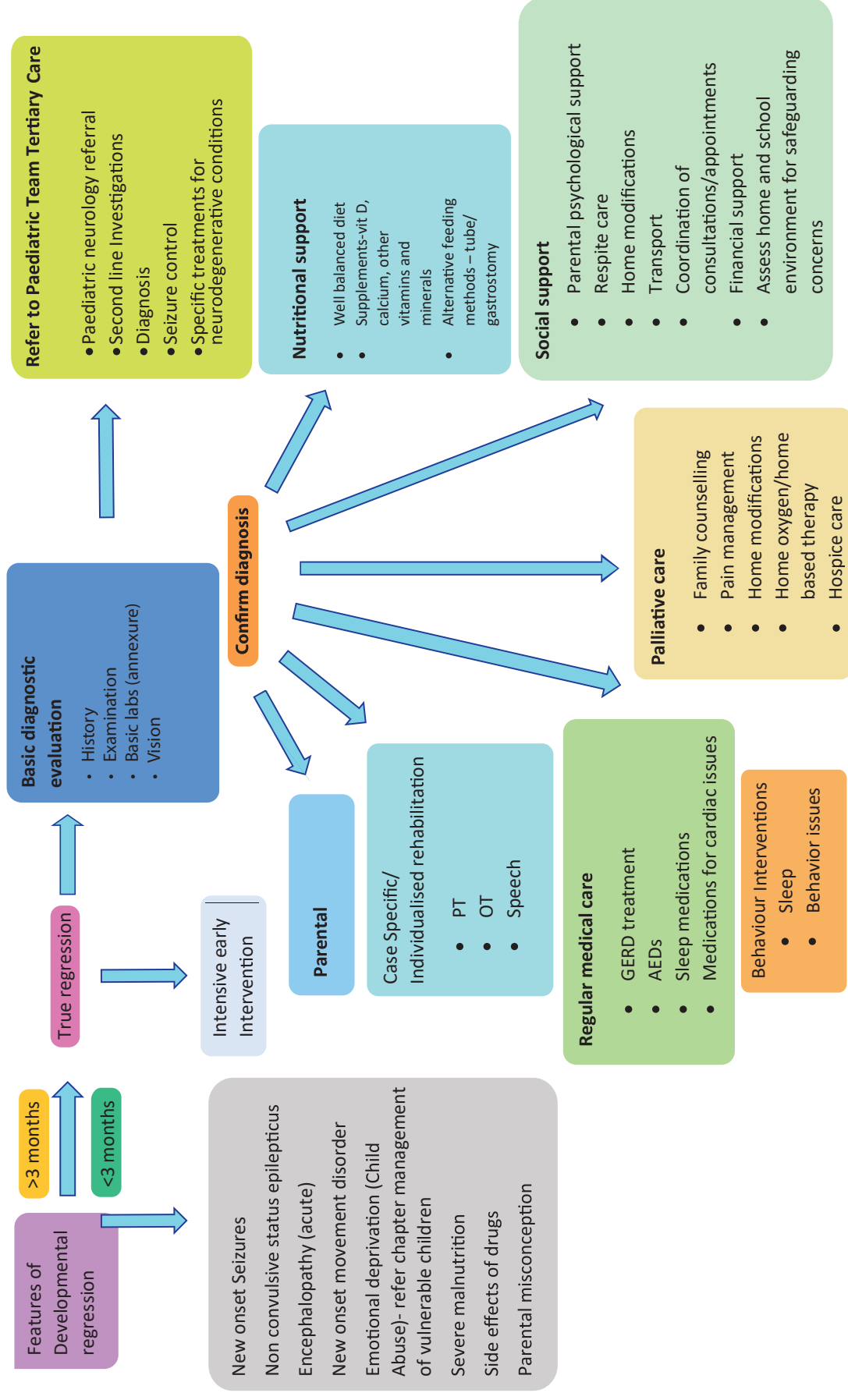


Figure 17: Management of a child with development regression

Functional assessment

Functional impairment can occur in all domains. Expected course is progressive decline, though plateaus of varying length can occur. No functional outcome measures have been standardized for Regressive disorders. Functional measures such as the Gross Motor Function Measure and Manual Ability Classification System may be utilized, keeping in mind that they are not validated for regressive disorders. Neuropsychological testing can also be useful in monitoring the progression of cognitive function. Communication assessment and provision of alternative augmentative communication (AAC) would help children and families.

Early predictions of outcomes

Diagnosis-dependent, but in general, the younger the onset, the more rapid the progression, and the worse the outcome.

Environmental

Evaluate the physical environment of the home and the school for accessibility and for safety and efficacy with performance of activities of daily living.

Social role and social support system

Explore whether patient and family have an adequate emotional support. Determine needs for financial assistance, respite care, monitoring and coordination of medical appointments and/or transportation. Explore patient's interest in recreational activities in order to develop adaptive programs as needed.

4.7 Rett syndrome

Rett syndrome (RS) is a neurodevelopmental disorder that occurs almost exclusively in females and has a typically degenerative course. Developmental potential for patients with Rett syndrome (RS) is difficult to predict. Some individuals with this syndrome achieve and maintain some functional skills. As many as 60% of RS patients may retain their abilities to ambulate; the remainder lose ambulation or never walk because of atrophy, dystonia, and scoliosis.

RS is related to various mutations on the MECP2 gene, which codes for methyl-CpG binding protein-2 (MECP2). Since the early post-mortem characterizations of brain abnormalities in Rett syndrome, later expanded by in vivo neuroimaging, there has been strong evidence for a generalized dendritic and synaptic disorder.

4.7.1 Management of Rett syndrome

A coordinated Multi-Disciplinary approach is preferred by a team consisting of General/Developmental / Community Paediatrician, Paediatric neurologist, Child psychiatrist, Nutritionist, Occupational therapist, PT, SLT, educational therapist and social worker. At an early age, shortly after the diagnosis, intensive early intervention as applied to other neurodevelopmental disorders is recommended.

Table 3: Management of Rett Syndrome

Management of Rett syndrome	Management of symptoms and health problems may require a multispecialty team. Regular monitoring of physical changes such as scoliosis and GI and heart problems is needed.
Medications	Though medications cannot cure Rett syndrome, they may help control some signs and symptoms associated with the disorder, such as seizures, muscle stiffness, or problems with breathing, sleep, the gastrointestinal tract or the heart.
Physical therapy	Physical therapy and the use of braces or casts can help children who have scoliosis or require hand or joint support. In some cases, physical therapy can also help maintain movement, create a proper sitting position, and improve walking skills, balance and flexibility. Assistive devices may be helpful.
Occupational therapy	Occupational therapy may improve purposeful use of the hands for activities such as dressing and feeding. If repetitive arm and hand movements are a problem, splints that restrict elbow or wrist motion may be helpful.
Speech-language therapy	Speech-language therapy can help improve a child's life by teaching nonverbal ways of communicating and helping with social interaction.
Nutritional support	Proper nutrition is extremely important for normal growth and for improved mental, physical and social abilities. A high-calorie, well-balanced diet may be recommended. Feeding strategies to prevent choking or vomiting are important. Some children and adults may need to be fed through a tube placed directly into the stomach (gastrostomy).
Behavioral intervention	Practicing and developing good sleep habits may be helpful for sleep disturbances.
Support Services	Academic, social and job-training services may help with integration into school, work and social activities. Special adaptations may make participation possible. Assess home and school environment for safeguarding concerns

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- Towards a better diagnosis and treatment of Rett syndrome: a model synaptic disorder-Abhishek Banerjee¹, Meghan T Miller²,Keji Li³, Mriganka Sur³, and Walter E Kaufmann
- Nelson book of Paediatrics-21st edition
- Fifteen-minute consultation: A practical approach to developmental regression in children
Thomas Smith, Dipak Ram

4.8 Acquired Brain Injury

4.8.1 Definition

Acquired brain injury (ABI) in children and young people is defined as brain insults acquired after the first life-year and includes both traumatic brain injuries (TBIs) and nontraumatic brain injuries (nTBIs) such as stroke, brain tumours, cerebral anoxia, and encephalitis.

ABI is the most common cause of death or disability among children and young people, with an incidence of 180 to 825 per 100 000 for TBI and 82.5 per 100 000 for nTBI in the world.

Severe ABIs are associated with impairments in physical, cognitive, and social-emotional function resulting in daily activity limitations, participation restrictions, and reduced overall quality of life and well-being.

Principles of rehabilitation

- Goal-directed, and evidence-based interventions
- Addresses the developmental stage and needs of the child
- Addresses physical, emotional, behavioural and cognitive rehabilitation needs of the child
- Is individualised and tailored to the needs of the child and their family/ carers and suit their home environments
- Directly involves “hands-on” and shared participation from parents, carers and other family members
- Adopts appropriate equipment

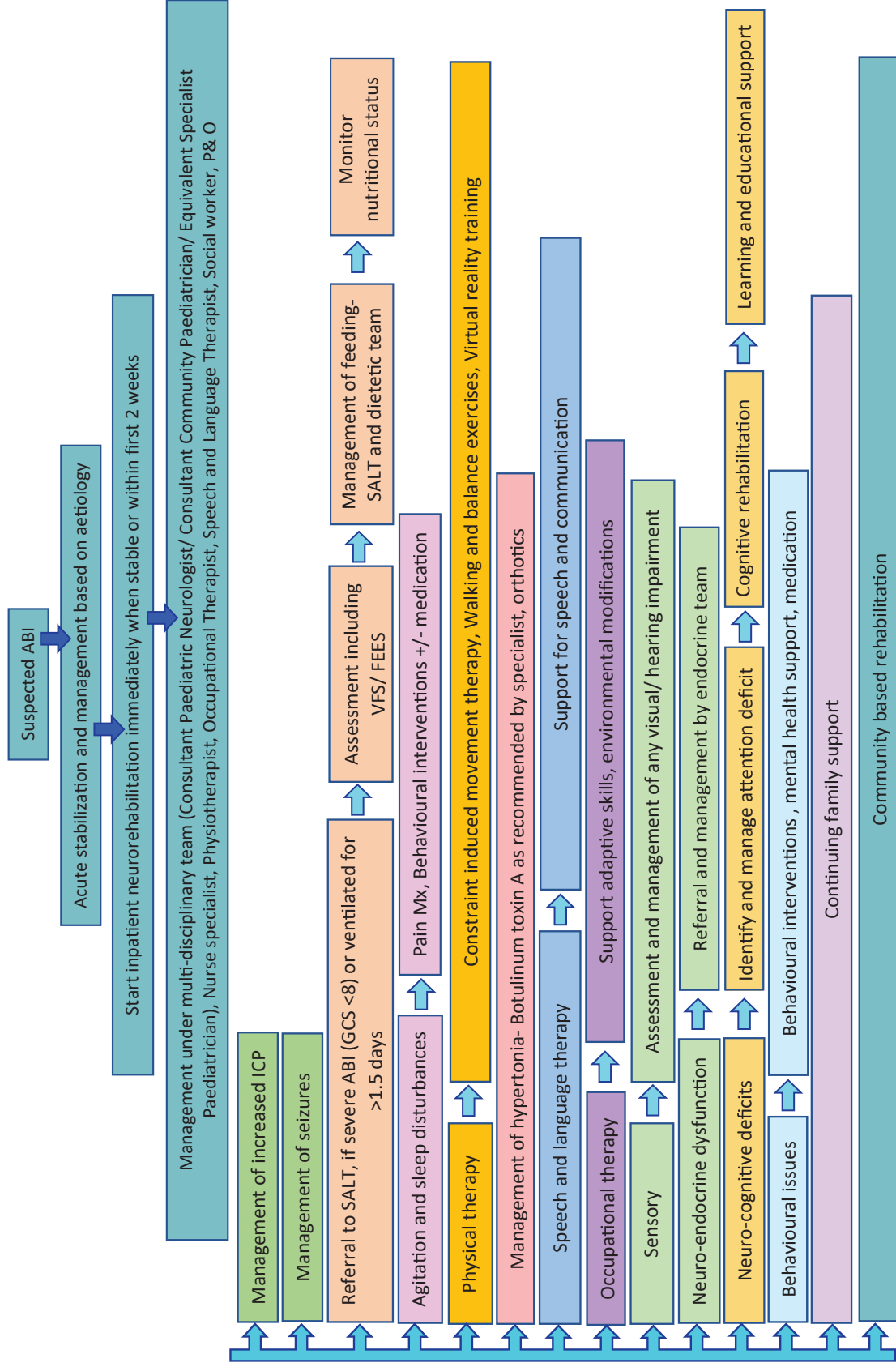


Figure 18: Management of acquired brain injury

Reference

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<https://www.sabin.scot.nhs.uk/wp-content/uploads/2018/06/Paediatric-ABI-Standards-April-2018.pdf>

4.9 Spinal dysraphism

Myelomeningocele (MMC) is the most common and most serious congenital anomaly of the human nervous system that is compatible with long term survival. It is associated with several comorbidities such as hydrocephalus, Chiari II malformation, tethered cord, scoliotic and kyphotic spinal deformities, skin injuries, as well as neurogenic bowel and bladder. **Each child who undergoes surgery should be referred for regular developmental follow up from the neuro surgical unit or the paediatric team.**

4.9.1 Clinical follow up

Table 4: Clinical follow-up of spinal dysraphism

Clinical follow up					
	Neonate	Infancy	Toddlers	Pre-schoolers	6-18 years
Follow up		6 monthly	6-12 months	6-12 months	12 months
Review the signs of acute shunt failure	headache, vomiting, and lethargy/sleepiness				
Review the signs of chronic shunt failure and review in clinic visits	accelerated head growth, loss of developmental milestones or neurological deterioration		low grade recurring headache and neck pain, loss of developmental milestones		recurring low grade headache and neck pain; loss of developmental milestones; cognitive, behavioral, neurological decline; urological regression
Review the signs of brain stem failure and review in clinic visits	poor control of oral secretions, swallowing dysfunction, stridor	poor control of oral secretions, swallowing dysfunction, stridor, and impaired language acquisition			poor control of secretions, swallowing dysfunction, stridor, and declining language function
Teach families the signs of TSC and review in clinic visits		back pain, declining lower extremity sensorimotor function			back pain, declining lower extremity sensorimotor function, bladder or bowel control decline and progressive orthopedic deformities, scoliosis
signs of syringomyelia			back pain, sensory changes in hands		
Investigations	Use supportive investigations judiciously to augment the clinical decision eg. MRI/CT, urodynamics, and sleep and swallow studies				

4.9.2 Mobility

Some individuals are able to ambulate with minimal impact from their Spina Bifida while others may require orthoses or assistive devices. In children and adults with Spina Bifida, the neurological level and motor impairment are the main factors that define an individual's potential for mobility.

Benefits of Mobility

- contracture management
- exercise: cardiovascular-respiratory effects
- strength effects and endurance
- community engagement/household mobility
- bone density
- bowel and bladder evacuation
- facilitates ability to perform self-care activities
- pressure reduction/redistribution
- Neurologic Level of Lesion and Anticipated Mobility

Mobility options according to level of NTD- Refer annexure

4.9.3 Orthopaedics

- Orthopaedic evaluations are recommended every three months in the first year of life.
- Consider hip imaging using ultrasound in the infant and anteroposterior pelvis radiographs after 6 months in patients with low lumbar or sacral lesions.
- Teach families about fractures and related precautions
- Perform spine evaluations by physical exam.
- Obtain scoliosis radiographs if a spinal deformity is suspected and monitor the progression.
- In children who have not achieved sitting balance, perform radiographs in a supine position. Once sitting balance is achieved, perform spinal radiographs in a sitting position. In children who can stand, perform in the standing position.
- Consider bracing or casting when there is a documented progression of scoliosis Monitor the spine for development or progression of a deformity that may be due to a tethered cord or syrinx.
- Monitor for deterioration of gait and consider treatment of orthopedic deformities leading to deterioration such as hip and knee contracture or rotational deformities.

4.9.4 Urology

Over 90% of children at birth have a normal upper urinary tract (kidney and ureter). Historically, we know that if left unattended, 50% of those children will suffer upper urinary tract damage due to lower urinary tract (bladder and urethra) hostility

Timely urology referral is essential.

4.9.4.1 Goals of management

- Maintain normal renal function throughout the lifespan
- Achieve urinary continence as early as socially acceptable
- Maximize urologic independence

- Reduce impact of urinary tract infections and antibiotic overuse.
- Establish a care program that allows for urologic independence, such as clean intermittent self-catheterization
- Determine the best measure of renal function
- Minimize occurrence of urolithiasis.
- Determine whether surgical interventions are effective in the long-term

4.9.4.2 Investigations

- Renal/bladder ultrasound - within three months and then repeat every six months till 2 year annually thereafter if the child is stable
- Urodynamic testing - within three months then yearly till 3 years
- Serum creatinine - within three months then repeat if upper tract changes are there or any other clinical indication, annually after 6 years

Initiate CIC and antimuscarinic therapy for the treatment of mixed incontinence when indicated based on the above results by a specialist.

4.9.5 Bowel Function and Care

Table 5: Bowel function and care of a child with spinal dysraphism

Infancy	• Monitor stool frequency, consistency, and amounts. • Use dietary management, in particular breastfeeding if possible, as it is easier to digest and offers better restoration of the microbiome after surgery • Consider dietary management (fiber and fluids) before pharmacologic adjuncts and/or rectal stimulants to manage constipation. • Use barrier creams to protect perineal area from breakdown as needed
Toddler Pre-schooler	• Discuss toilet training and habit training with parents. • Establish goal of working toward bowel continence. • Focus on timed bowel movements after meals. • Use dietary management (fiber and fluids), pharmacologic adjuncts and/or rectal stimulants to manage constipation and fecal incontinence. • Use barrier creams to protect perineal area from breakdown as needed
6-18 years	• Discuss consequences of constipation and bowel incontinence (including shunt malfunction, urinary incontinence, UTIs, skin breakdown, social isolation) • Focus on developing independent management skills • Assist the child with learning how to minimize and manage bowel accidents. • Use barrier creams to protect perineal area from breakdown as needed • Keep a bowel habit diary to better understand triggers for incontinence and identify a choice of options for bowel management. • Focus on fiber, fluids, exercise, and timed bowel movements after meals. • Pharmacologic adjuncts to manage constipation

4.9.6 Nutrition, Metabolic Syndrome, and Obesity

Table 6: Nutritional management of a child with spinal dysraphism

Infancy	- Assess weight, height and occipital frontal circumference at every clinical encounter. - Support on breastfeeding and encourage mothers to nurse their child in a flat position for five days following surgery to reduce pressure on the wound and avoid a cerebral spinal fluid leak - Mention that severe Chiari malformation may affect attachment and coordination of sucking, swallowing, and breathing.
Toddler-Pre-schooler	- Conduct annual assessments of weight, height or arm span, and calculate BMI. BMI is an imperfect indicator of health in children with Spina Bifida due to difficulties measuring height. Consider monitoring other measures of adiposity, such as waist circumference. - Educate families on the importance of consuming a balanced diet and how it affects the whole body. - Discuss that poor eating habits and reduced activity may lead to obesity, constipation, skin breakdown, osteoporosis, anaemia, and other problems - Highlight that children with Spina Bifida, especially those who are non-ambulatory/ low levels of physical activity, and who have higher body fat levels or contractures , are at increased risk for fractures. Recommend a diet with adequate calcium and vitamin D.
6-18 year	- Conduct annual assessment of weight, height or arm span, and BMI, waist circumference, blood pressure. - Discuss the importance of consuming fiber and water to manage bowel and bladder health . - Suggest parents to let their children choose a new healthy food to try. - Involve children in discussions about healthy lifestyles in order to explore their understanding, perceptions, and priorities regarding nutrition .

4.9.7 NCD Screening

Screening for dyslipidemia is recommended every two years from two years of age if the child's BMI is above 95th percentile or a family history of dyslipidemia/early cardiovascular disease and/or morbidity in first- or second-degree relatives is present

- Screening for diabetes is recommended every two years in children 10 years of age /puberty and for all children with a BMI over the ≥ 85 th percentile and who have two or more additional risk factors including
 - family history of type 2 diabetes mellitus/ acanthosis nigricans / hypertension/ dyslipidemia / polycystic ovary syndrome

4.9.8 Skin Care

- Teach parents and caregivers to inspect the skin daily for changes in colour, texture, and temperature and advice to keep skin clean and dry.
- Encourage the child's involvement in skin inspection and teach child to develop awareness of insensate areas.
- Teach parents and caregivers that the child should wear protective clothing and footwear over insensate areas.
- Promote adequate hydration and proper nutrition for healthy skin.
- Suggest children and adults who use wheelchairs to use a pressure-relieving cushion and check it daily.
- Identify and discuss risk factors of pressure injuries, such as using a wheelchair, having had surgery above the knee, shunts, a higher level of lesion, recent surgery, bladder incontinence, and being of the male gender.
- Advise children and adults who are non-ambulatory and use a wheelchair to engage in pressure-relieving activities every 15 minutes and teach safe transfer skills to non-ambulatory patients.
- Tell parents and caregivers to seek treatment if the child's skin is compromised.

4.9.9 Important Points in Counselling

Provide information about folic acid and discuss recurrence risk and potential impact on future pregnancies.

Reassure the parents that while NTDs are not completely understood, this birth defect was not something that was caused by their actions.

Reassure the parents that while folic acid can help diminish the chance of a pregnancy being affected by NTD, it is not entirely preventative. Counsel women about their recurrence risk and promote daily consumption of 4 milligrams (4000 micrograms) of folic acid beginning at least one month (but preferably 3 months) before they start trying to get pregnant and continuing through the first three months of pregnancy.

4.10 Visual impairment in children

An estimated 19 million of children are visually impaired worldwide, while 1.4 million are blind¹. Though visual impairment is less common in children compared to adult population, the impact of it is very large and lifelong especially in psychological, educational and socioeconomic aspects.

4.10.1 Defining visual impairment

Most systems for classifying Visual Impairment are based on acuity in the better eye and to a lesser degree the visual field. A child with impaired vision (of any severity) in only one eye due to unilateral / asymmetric disease is not formally considered ‘visually impaired’. The WHO categorization system for visual impairment has been very widely but not universally adopted.

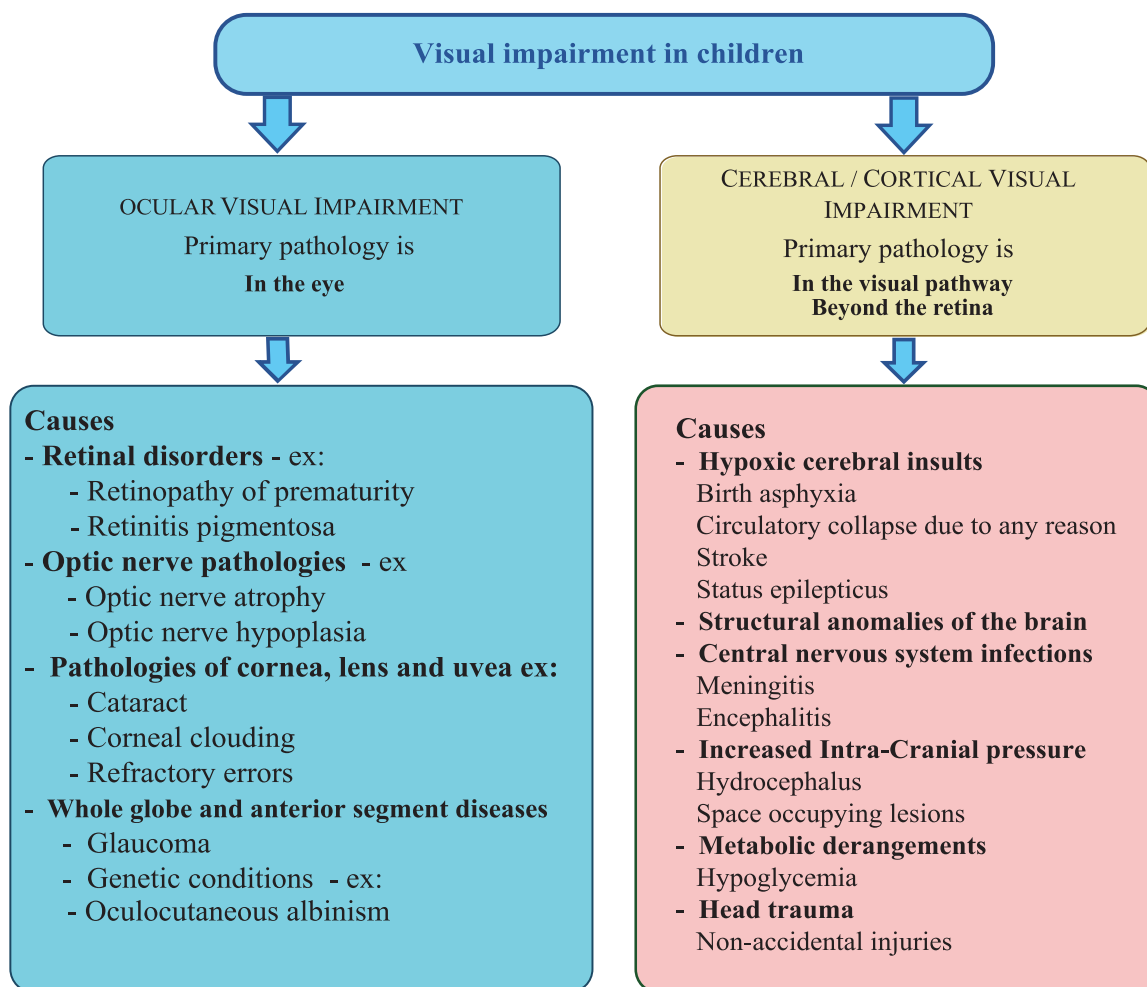


Figure 19: Aetiology of visual impairment

4.10.2 Evaluation of a child with visual impairment

Modes Of Presentation

Newborn assessment
Routine health supervision check ups
Caregiver concerns

- Poor eye contact
- Poor social interaction
- Delayed achievement of developmental milestones

Concerns by school teachers

- Need of sitting near the board
- Need of large fonts

Screening due to high risk conditions / illness
Incidental finding while being evaluated for another illness

History

Complaints from the caregiver
Suggestive of ocular visual impairment

- My child has no/poor eye contact
- My child is extremely sensitive to light
- My child's eye looks different in size / alignment
- My child doesn't smile at me
- My child sees only the nearby objects

- 1. Antenatal**
 - Maternal immunization failure
 - Congenital infections
 - Ultrasonically detected congenital abnormalities
- 2. Perinatal**
 - Prematurity
 - Intra Uterine Growth Retardation
 - Low APGAR
 - Need of resuscitation
- 3. Postnatal**
 - Stay at SCBU
 - Need of respiratory support
 - Infections - sepsis, meningitis
- 4. Developmental history**
 - Delays / regression in any of the developmental domains
- 5. Past history**
 - Acute illnesses
 - ICU stays
 - Trauma
 - Surgeries
- 6. Family history**
 - Blindness
 - Genetic disorders
- 7. Social history**
 - Impact on activities of daily living
 - Impairment of scholastic performances

Complaints from the caregiver
Suggestive of cerebral/cortical visual impairment

- My child has no/poor eye contact
- My child is staring at light
- My child's eye looks different in size / alignment
- My child doesn't smile at me
- My child sees only the nearby objects
- My child enjoys only at moving objects
- My child looks only bright colored objects
- My child looks only at illuminated toys / toys make sound

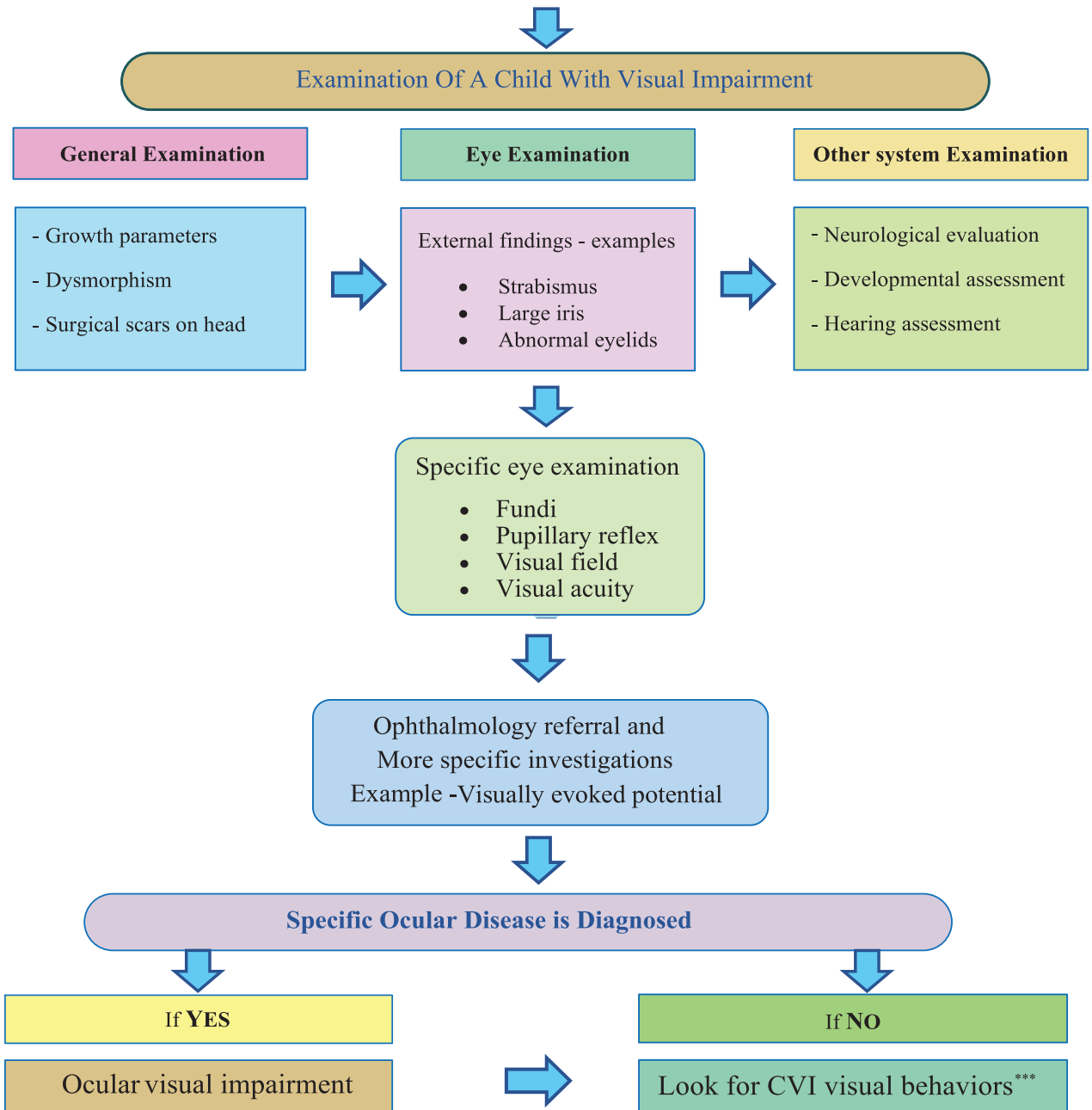


Figure 20: Evaluation of a child with visual impairment

4.10.3 Red flags for visual development

The visual milestones may not be achieved due to motor cognitive or other difficulties.
The milestones in bold letters are mostly independent of above.

At Any age

- **No response to visual threat**
- **No blink response to tap on forehead**
- **Gazing at lights/ no objections to a bright light shone in the eye**
- **Closing eyes in an environment with normal background light/ rubbing eyes after looking at an object observed repetitively**
- **Constant absence of fixing at a bright object at 8-12 inches**
- **Looks only at objects with 1 or 2 favorite colours**
- Abnormal head posture when looking
- **Abnormal eye movements/ nystagmus**
- **Squint/ dyssyugate eye movements beyond 5 months**

Table 7: Red flags for visual development according to the age

Age	Sign
Birth to -1 month	• Infant blink or close eyes in response to bright light or touching eye • Able to stare at object if held 8 to 12 inches away • Fixes eyes on a face or light • May follow a moving object
1 -2 months	• Looks at faces and pictures with contrasting black and white images • Can follow an object up to 90 degrees • Watches parent closely • Smiles at caregiver consistently as a response to seeing (when silently approached) by 2 months
2 to 3 months	• (Hand regard) Is baby aware of his/her hands • Look directly at parent's or caregiver's eyes • Follows beyond 90 degrees • Smiling at his/her parent or caregiver without need for attracting with sound
4 to 5 months	• Beginning to reach hands to objects, faces • Recognizes familiar objects such as toy/ bottle (gets excited) • Shifts eyes from one person to another • Looks with eyes together by 5 months
5 to 7 months	• Consistently reach for objects • Will turn head to see an object • Follows objects when placed near (< 1 foot) and far (> 6 feet) by 7 months
7 to 11 months	• Recognize family members and/or caregivers and have a welcoming smile before hearing voices or seeing his/her smiles? • look at small objects, such as rice grains /moving insects cereal or a raisin • Plays pee k-a-boo
12 to 14 months	• Able to place shapes in proper holes • Becomes interested in pictures • Recognizes familiar objects and pictures in books, may point to some objects when asked, "Where is the ...?" • Points and gestures for objects • Copies actions

Management Of Ocular Visual Impairment

Ophthalmology team	Follow up	Additional supports for
- Disease specific management ex - surgery, glasses	- Regular visual examination - Visual rehabilitation - Developmental assessment	- Activities of daily living - Scholastic function - Parental empowerment

Specific Ocular Disease Is Diagnosed Or Not



Look for CVI visual behavior



Visual and behavioral characteristics of Cortical Visual Impairment***

1. Strong color preference - especially for red / yellow
2. Visual field preference - the presence of unusual field locations in addition to loss of visual field
3. Need for movement to elicit / sustain visual attention
 - Either the viewer or the object viewed needs to be moving to maximize the viewer's ability to view the object
4. Visual latency - delayed responses in looking at objects
5. Difficulties with visual complexity -
 - Object itself may present a complex display
 - Object is viewed within a complex environment
 - Another sensory input is competing for the viewer's attention when an object is viewed
6. Light gazing and non-purposeful gazing
7. Difficulty with distance viewing
8. Difficulty with visual novelty - a preference for seeing familiar objects
9. Absence of visually guided reach (atypical pattern of visual - motor behavior)
 - The ability to look at and touch an object at the same time is not displayed
 - These actions are performed separately
10. Absent / atypical visual reflexes
 - the reflex to blink in response to an approaching object is impaired

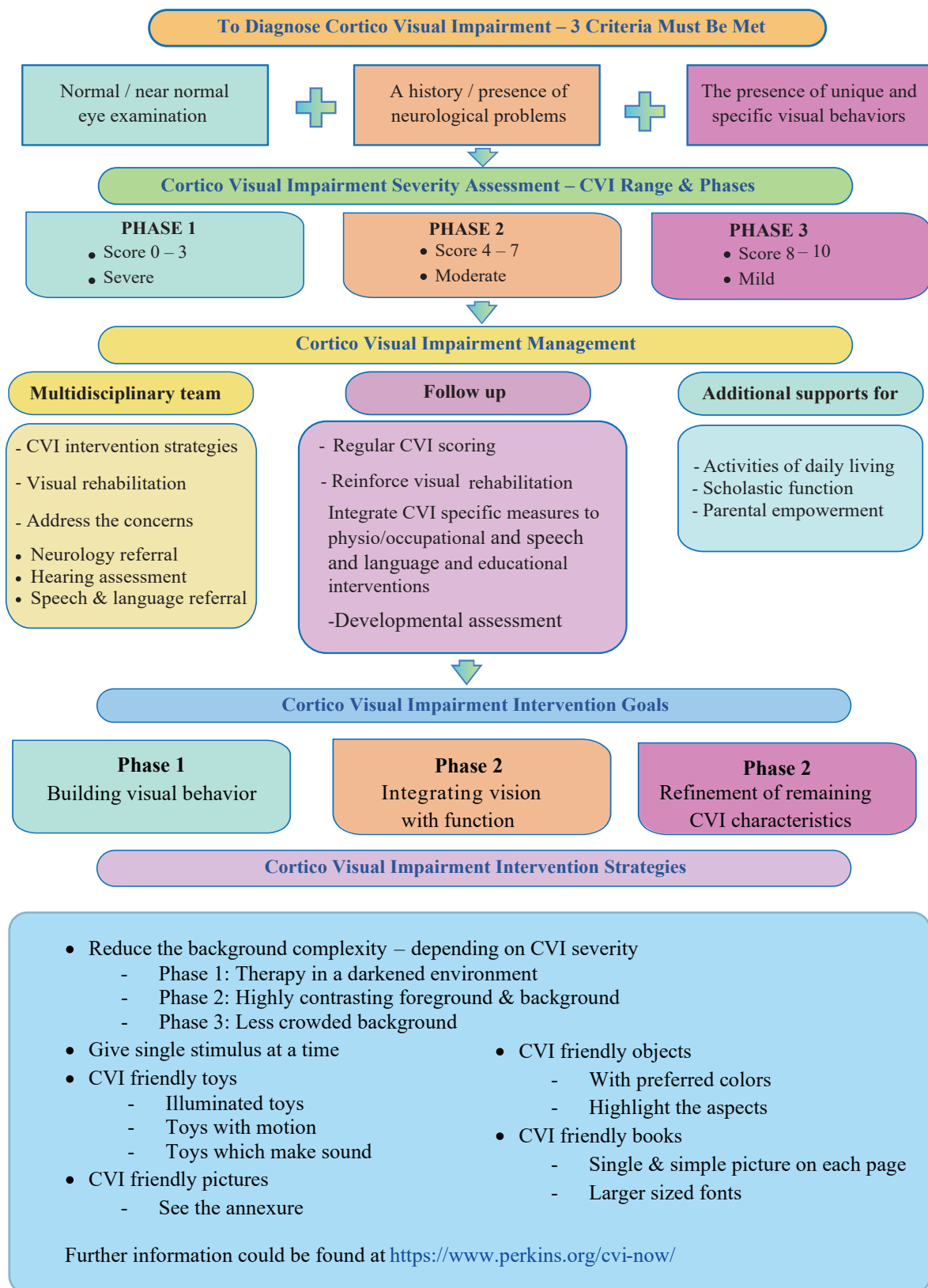


Figure 21: Management of ocular visual impairment

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Recent advances in Cortical Visual Impairment – Developmental Medicine and Child Neurology
2001 – USA

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5. Approach to a child with suspected developmental disability

may present with unidentified delay in development. These may include, but not limited to unidentified syndromes, genetic mutations, metabolic disorders, infections, undiagnosed neuro developmental disorders (ie : ASD, ADHD), primary hearing/ vision impairment and under stimulation

Always take a developmental history in all children who present to you

Definitions

Development delay

A significant delay (deficit in performance off at least 2SD below the age appropriate mean on accepted standardized tests) in two or more developmental domains affecting children under the age of five years is termed global developmental delay.

Global Developmental Delay (GDD)

Defined as a delay in two or more developmental domains affecting children under the age of 5 years.

Remember,

Dysmorphic features, delay in achieving development milestones could have been overlooked.

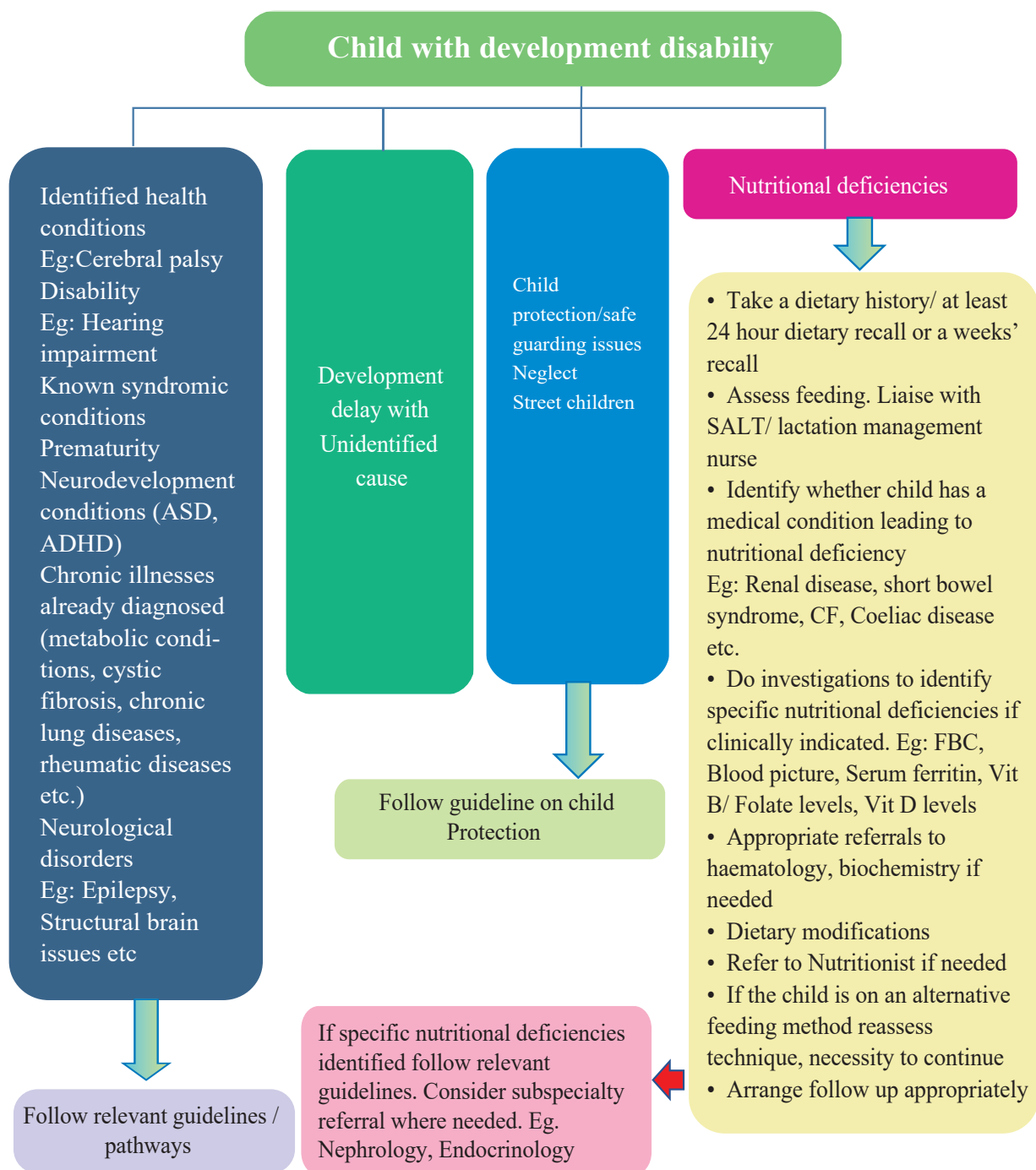
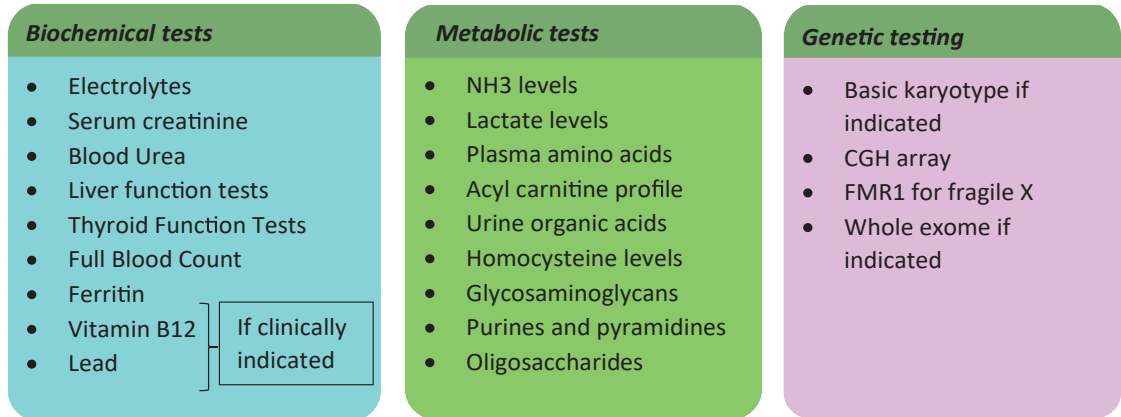


Figure 22: Approach to a child with suspected developmental disability

5.1 Children with a development delay with an unidentified cause

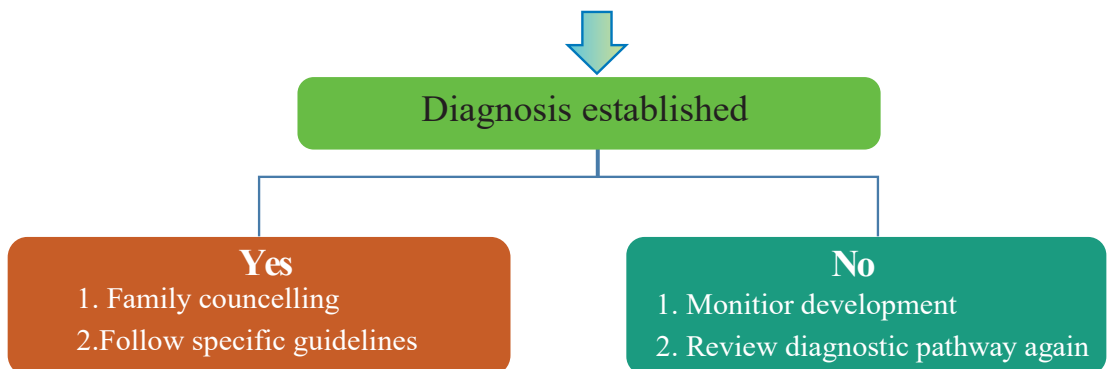
1. Do a thorough development assessment. Record with a development assessment tool. Eg. Bayley, Griffiths
2. Hearing/ vision checks
3. Assess for learning difficulty
4. Investigate as needed



5. Refer to clinical geneticist as needed
6. Second line investigations if needed

-
1. Neuroimaging - MRI preferred
 2. Neurophysiology
 - EEG - For uncontrolled Epilepsy or Epileptic Syndromes, Speech regression, or neurodegenerative disorders
 3. Testing for congenital infections (TORCH Screen) when with neurological anomalies, microcephaly, hearing and/or vision loss.

7. Meantime involve PT/ OT/ SLT Multidisciplinary approach



8. Identify whether these children have under stimulation for development. Assess home environment.
9. Children with development disability have higher risk of neglect and maltreatment. When assessing children always have this in mind and be alert to any signs of child maltreatment. Always assess home and school (when appropriate) environment, for considerations, such as abuse/neglect/maltreatment/exposure to violence in the family, lack of stimulation and priority mental health conditions in caregivers (e.g., maternal depression), etc. Always look for warning signs like explained bruises, pain, weight loss and behavior changes (including avoidance of certain people or places) that may indicate victimization. Some behavioral changes to keep an eye out for are social withdrawal, avoidance of particular places or people, behavioral outbursts when presented with particular places or people, or developmental regression. *for further details please follow National Guideline for The Management of Child Abuse and Neglect

5.2 Lack of stimulation

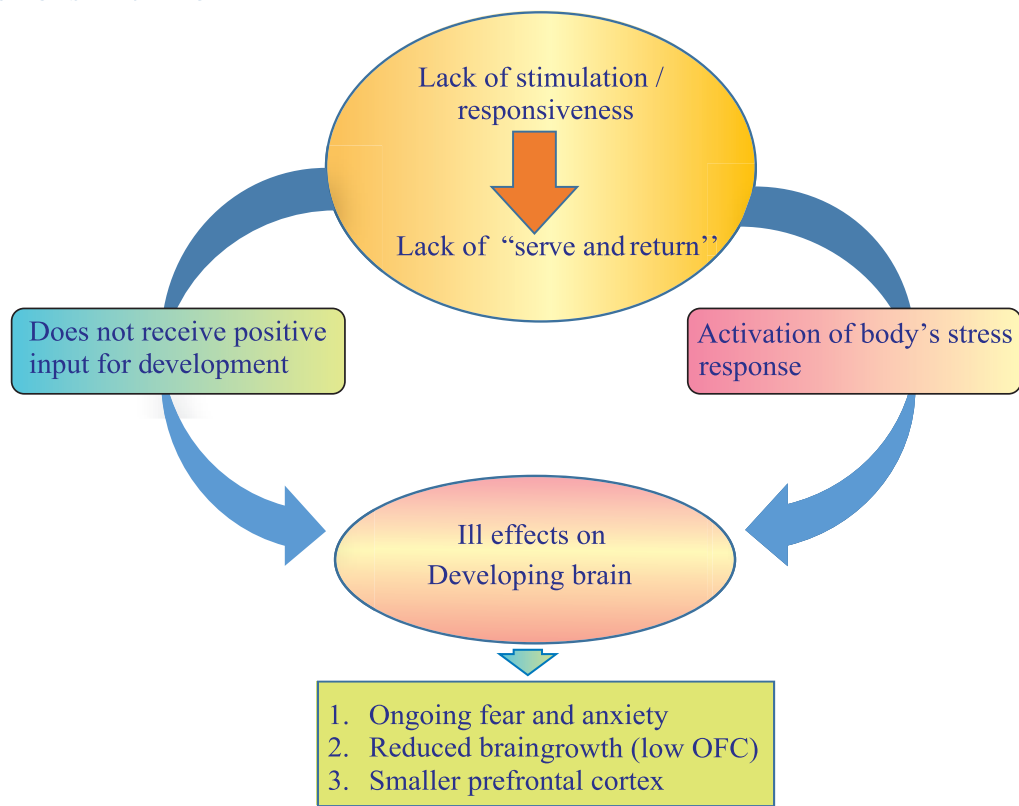


Figure 23: Lack of stimulation / responsiveness

5.3 Addressing lack of stimulation

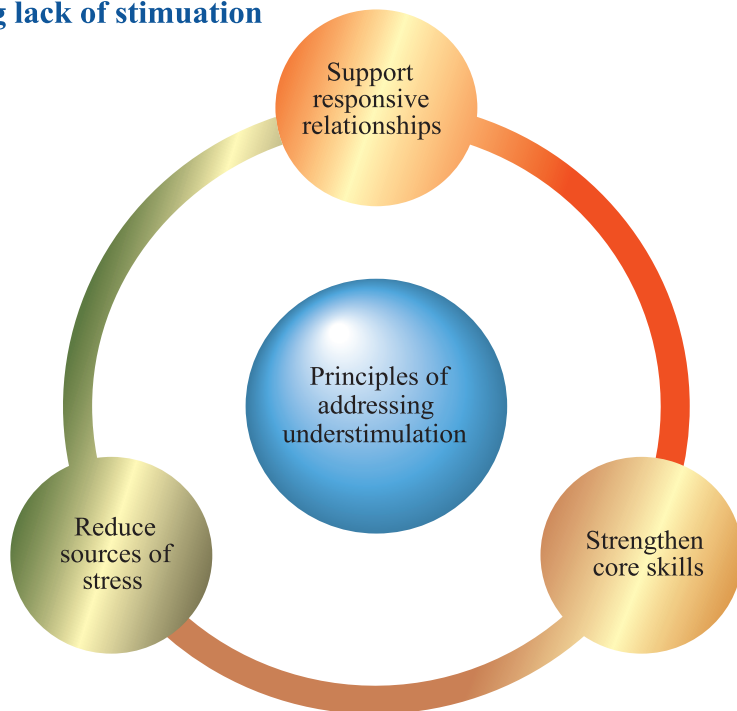


Figure 24: Addressing lack of stimulation

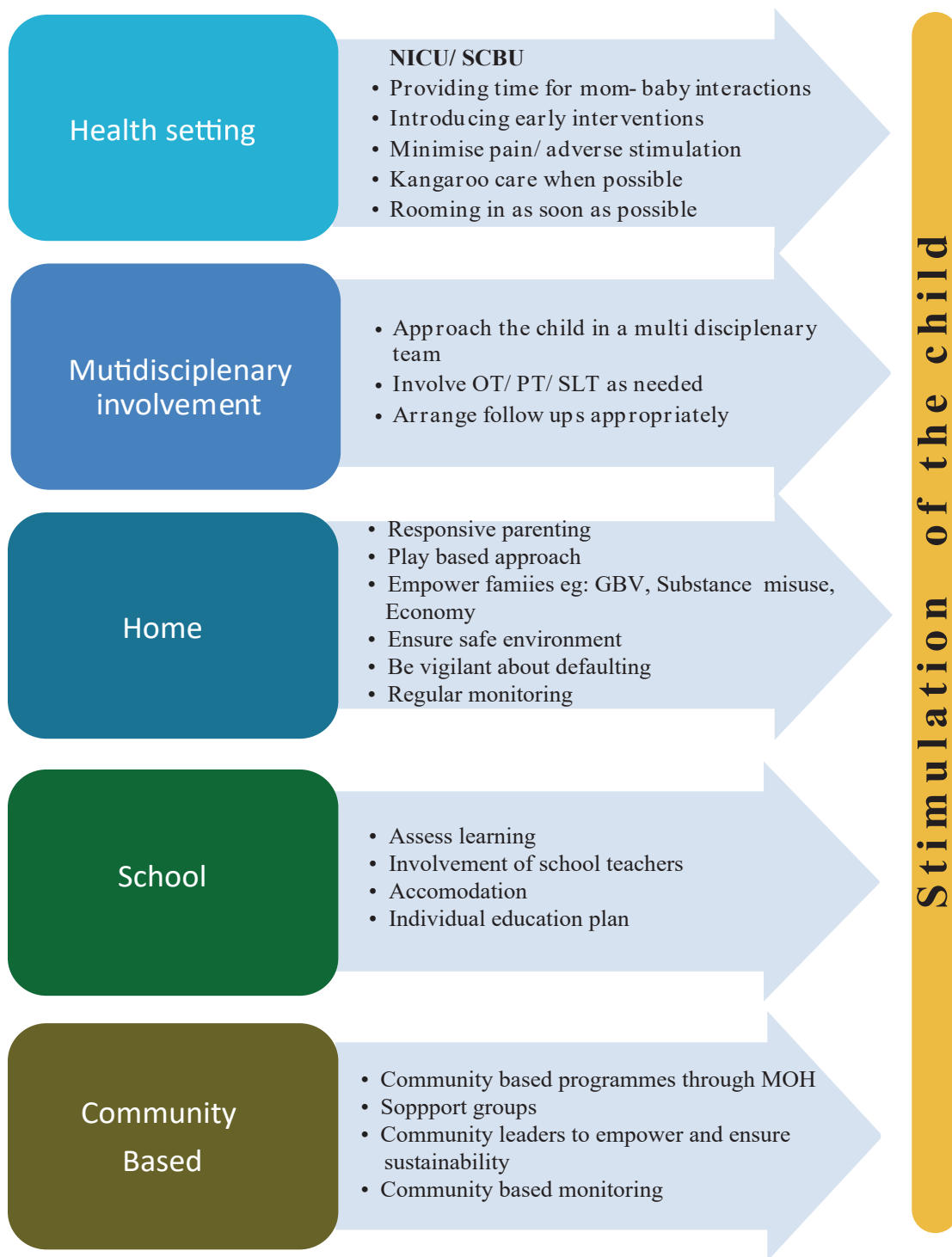


Figure 25: Stimulation of the child in different settings

6 Risk factors for child maltreatment

Studies have shown that children with disability have at least 3 times more risk of maltreatment compared to their peers without disability. Therefore, you should be very vigilant about presence of such signs indicating child maltreatment in all children you come across with disability.

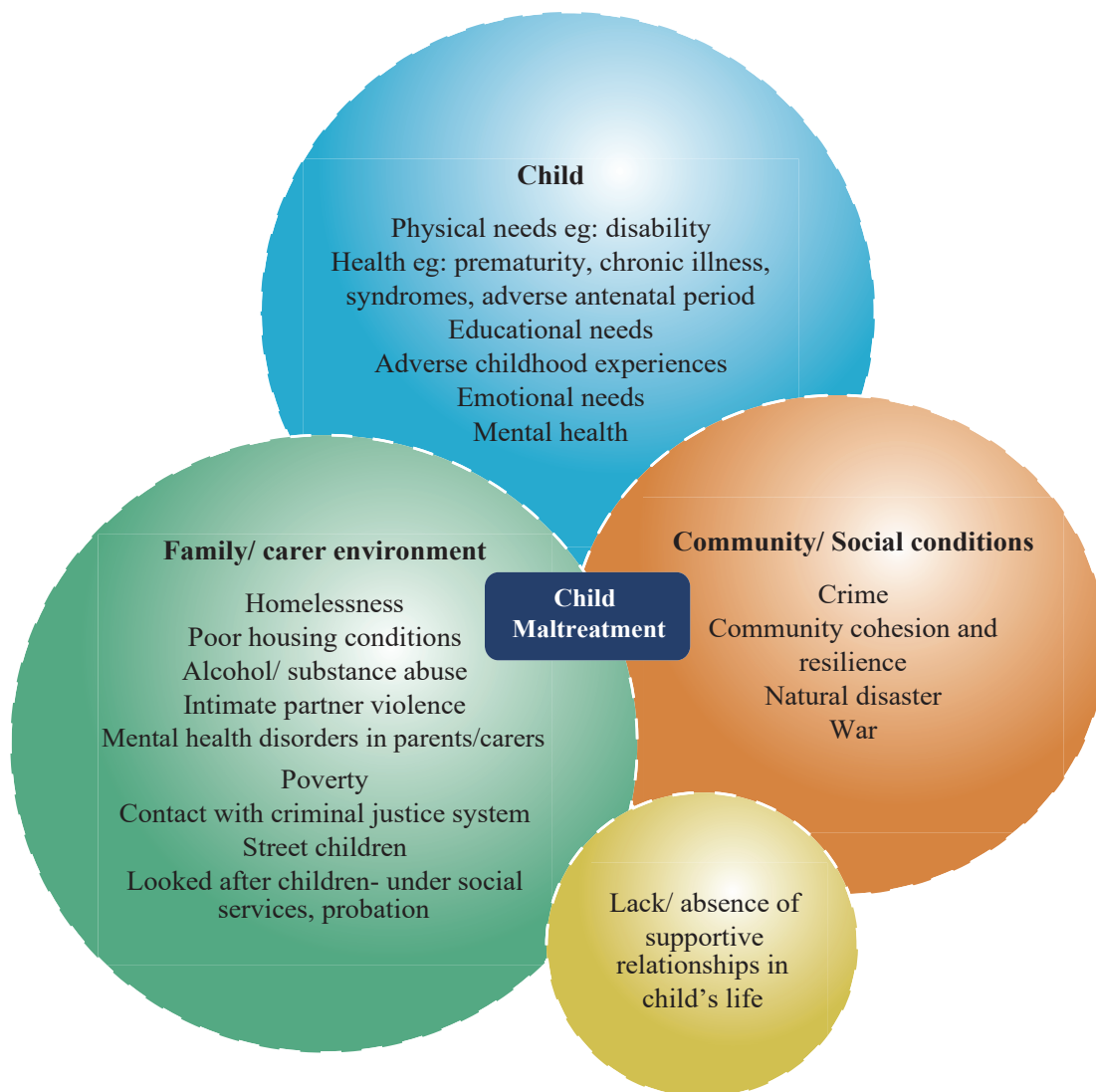


Figure 26: Risk factors for child maltreatment

Most of the time these factors are interrelated and will create a vicious cycle

Presentation

- Nutritional deficiencies
- Delay in development
- Child protection/safeguarding issues
- Chronic illness
- Challenging behavior
- Learning difficulties
- Mental health issues
- Metabolic disorders
- Self-harm

Identify risk factors/ lack of stimulation

1. Take a good history including social history and developmental/ schoolwork history
2. Examine. Be vigilant to identify features of neglect, deprivation.
Eg: (This list is not exhaustive)
 - General appearance
 - Syndromic features
 - Personal hygiene (nails, dress, in nappies or not)
 - Dental caries
 - Nutritional deficiencies (always complete anthropometry, make a note of hair/ nails)
 - Features suggestive of child abuse (scars, fractures, features of neglect etc)
3. Make a note of child- parent/ carer relationship/ bond
4. Do a screening development assessment in children less than 6 years

Follow assessment pathways for suspected development delay if present.
Follow guidelines for child protection if maltreatment suspected

Figure 27: Evaluation of child maltreatment

Annexure 1

7.1 Tips for parents to improve parenting skills**1. Celebrate the Positive**

Recognizing children for their effort is as important as the actual effort

2. Take Time to Talk and Really Listen

Let children know talking through their feelings is a healthy way of expressing themselves and parents need to be a role model by talking about your feelings

3. Use Discipline, Not Punishment

A punishment force child to behave a certain way but discipline teaches lifelong responsibility. Use natural and logical consequences to teach them lifelong responsibility and learning.

For example: if a child naughty with others rather than punishing him discuss what are the nice ways to deal with others and appreciate when he does so.

4. Teach responsibility constructively

Opportunities to practice making decision

For example, instead of saying “You made a mess, you left your dishes on the table and you need to take them to the sink and rinse them.” Try saying, “The table is a mess, what do you need to do to get it cleaned up?”

5. Separate the Child from the Behaviour

Never tell a child that he/she is bad. Communicate with the child that you don’t tolerate his/her bad behavior but not him/her. You should show them that your love is unconditional

For example, instead of saying, “You are acting like a baby,” try saying, “I know you are upset but I can understand you better if talk to me in your regular voice.”

6. Actions speak louder than words

Stop giving commands again and again.

For example, if your child continues to throw his towel on the bathroom floor rather than hang it up, instead of doing it for him or yelling at him one more time, simply remove the towel or leave it in a heap on the floor. It will either be gone or still damp the next time and will better deliver the message of why it is important to hang it up

7. Use Natural and Logical Consequences

Give children a chance to learn from the consequences of their actions

For example, even though he has to complete his home works gradually during the vacation, instead of doing that if he does it within the whole last day of the vacation. You just sign there, by mentioning his actual action and let him deal with consequences from the teacher.

8. Spend quality time with your child and leave the stress of work at work

In a child's mind, negative attention is better than no attention at all.

Separate a time which is dedicated for children from your tight schedule and that should be totally for them even though it is a short duration

Parents are all busy, but it is important to remember not to take your job home with you.

9. Give children input into the decisions including family decisions at the time

Ask your child's advice when it is appropriate to do so. This can help a child feel powerful and valuable. Give your child choices, let him/her help you, and have input into simple daily decisions.

10. Be kind although firm and consistent (by all the family members)

Finding positive ways to talk about your children	
Instead of.....	Say.....
"She talks too much."	"She is very verbal."
"He asks too many questions."	"He is curious about many things."
"He tries to dominate everything."	"He shows leadership abilities."
"He is argumentative."	"He is good at defending his position."
"She always looks for the simple solution."	"She is good at solving problems."
"He is stubborn."	"He is goal directed."

Promoting Responsibility

Use this guide to help teach children to think about their actions and make decisions for themselves

Handling typical situations with "think starters" like the suggestions noted in the chart below will encourage children to be responsible for their own behavior.

Situation The parent is on the phone or busy with another child, other child is noisy	Think stopper “Be quiet. You’re making too much noise.”	Think starter “Your noise is making it hard for us to hear. What can you do to help?”
It’s clean-up time.	“Put the dishes in the sink, pick up the toys in the living room, and wipe off the tables.”	“It’s time to clean up. What do you need to do to clean up?”
One child is distracting another.	“Daham, move away from Dulara.”	“Daham, find something else to do so that Dulara can concentrate”
One child is making noise while another is trying to speak.	“Be quiet.”	“It’s Minidu’s time to talk. What do you need to do?”
Siblings are fighting.	“Stop it! Each of you sit on opposite sides of the room.”	“We can’t have fighting. Take time to cool off and then let’s talk about what you can do to let someone know that you are angry with him.”
A child is scared of the dark.	“There is nothing to be afraid of.”	“I know you are scared when your room is dark. What would make you feel more comfortable?”
A child is upset about a grade on a test.	“You should have studied harder.”	“What are you going to do to improve your grade?”
A child can’t figure out how to do something and says, “I can’t do this.	“Here’s how to do it: First you.”	“Which part is giving you trouble

7.2 Possible aetiology for developmental impairment

Broad category	Possible causes
Prenatal intrinsic	Genetic Central nervous system malformations Metabolic
Prenatal extrinsic	Teratogens/toxins (drugs of abuse, medications, etc.) Infections
Perinatal	Perinatal hypoxia Prematurity Neonatal complications
Postnatal	Neglect/psychosocial environment Infections Trauma Toxins

7.3 Commonly seen dysmorphic features and their definitions

Feature	Definition
Brachycephaly	head shape is shortened along the sagittal plane
Plagiocephaly	head shape is asymmetric in the sagittal or coronal plane
Scaphocephaly	head is elongated from front to back in the sagittal plane
Brachydactyly	Short digits
Camptodactyly	Permanent flexion of one or more fingers associated with missing inner phalangeal creases
Clinodactyly	A medial or lateral curving of the fingers; usually refers to incurving of the 5 th finger
Postaxial Polydactyly	Extra finger or toe present on the lateral side of the hand or foot
Preaxial polydactyly	Extra finger or toe present on the medial side of the hand or foot
Syndactyly	Incomplete separation of the fingers. It most commonly occurs between the 3 rd and 4 th fingers and between the 2 nd and 3 rd toes.
Hypoplastic nail	An unusually small nail on a digit
Low-set ears	Helix meets the cranium at a level below a horizontal plane drawn through inner canthi of eyes
Ocular Hypertelorism	Increased distance between the pupils of the two eyes
Short palpebral fissures	Decreased horizontal distance of the eye based on measurement from the inner to the outer canthus

Some common syndromes with genetic basis, their features and management principles

Syndrome	Clinical features	Management
Fragile X syndrome	Macrocephaly in early childhood, accelerated linear growth in childhood. Mild to profound intellectual disability. Communication and language needs, Autism, hyperactivity, Sensitivity to stimuli, leading to serious behaviour problems in overstimulating situations. Prominent forehead, elongated face, prognathism usually not noted until after puberty, thickening of nasal bridge extending down to the nasal tip, large ears with soft cartilage, epicanthal folds, high arched palate, dental crowding.	Speech and language therapy Learning and behavioural support
Turner syndrome	Small stature, delays in motor skills, poor coordination, visual-spatial organization deficits, ovarian dysgenesis with hypoplasia, congenital lymphedema, broad chest with widely spaced nipples, low posterior hairline, appearance of short neck, webbed posterior neck, cubitus valgus, dislocation of hip, horseshoe kidney, double or cleft renal pelvis, bicuspid aortic valve, coarctation of aorta, valvular aortic stenosis, mitral valve prolapse, hearing impairment	Endocrine referral Follow up for renal anomalies Cardiac referral Learning and behavioural support

22q11.2 deletion DiGeorge syndrome Velocardiofacial syndrome	Cardiac defects especially outflow tract, Cleft palate, micrognathia, prominent nose, overturned helix of ear, hypocalcaemia, absent thymus	Cardiac referral Endocrine referral OMF referral Feeding support Learning and behavioural support
Prader-Willi syndrome	Neonatal hypotonia Bitemporal narrowing, Almond-shaped eyes, Tube feeding required	Endocrine referral Feeding support Learning and behavioural support
Angelman syndrome	Severe intellectual disability, marked delay in attainment of motor milestones, movement or balance disorder, poor language development/non-verbal, frequent laughing/smiling, apparent happy demeanor, easily excitable personality often with uplifted hand-flapping, microbrachycephaly; decreased pigmentation of the choroid and iris, maxillary hypoplasia, deep-set eyes, a large mouth with tongue protrusion and widely spaced teeth; prognathia, ataxia and jerky arm movements resembling a puppet gait, drooling; excessive chewing/mouthing behavior; seizures varying from major motor to akinetic, beginning usually between 18 and 24 months, EEG abnormalities, hypotonia, cerebral atrophy	Speech and language therapy Neurology referral Physical therapy Learning and behavioural support
Marfan syndrome	Long limbs, arachnodactyly, contractures, enophthalmos, dislocated lenses, wrinkly skin, heart murmur family history of Marfan syndrome	Cardiac referral Ophthalmology referral Physical therapy Learning and behavioural support
Pierre Robin sequence	Cleft palate and micrognathia. Flat nasal bridge, prominent eyes, joint laxity Eye examination shows myopia and vitreous abnormalities (not often apparent at birth). Mild platyspondyly on spinal X-ray.	OMF referral Feeding support Eye referral
Noonan syndrome	Short stature of postnatal onset, epicanthal folds; ptosis of eyelids; hypertelorism. low nasal bridge; down slanting palpebral fissures; myopia; keratoconus; strabismus. nystagmus; low-set or abnormal auricles. prominent, protruding upper lip; moderate retrognathia, low posterior hairline, short or webbed neck, shield chest and pectus excavatum or pectus carinatum, cubitus	Cardiac referral Ophthalmology referral Physical therapy Learning and behavioural support

	valgus, scoliosis with thoracic lordosis, abnormalities of vertebral column, pulmonary valve stenosis, hypertrophic cardiomyopathy, atrial septal defect, tetralogy of Fallot, small penis, cryptorchidism, defects in the coagulation and platelet systems, thrombocytopenia	
William syndrome	Mild microcephaly, Average IQ, friendly personality, mild spasticity, poor coordination, hypotonia, short palpebral fissures, depressed nasal bridge, epicanthal folds, periorbital fullness, anteverted nares, long philtrum, prominent lips with open mouth, supraaortic stenosis, pulmonic valvular stenosis, renal artery stenosis with hypertension, hypoplasia of the aorta, and other arterial anomalies, microdontia, joint hypermobility, contractures, lordosis, scoliosis, kyphosis, renal anomalies, including nephrocalcinosis, bladder diverticula, urethral stenosis, vesicoureteral reflux	Cardiac referral Nephrology referral Ophthalmology referral ENT referral Physical therapy
Rubinstein-Taybi syndrome	Broad medially deviated thumbs and great toes, short columella, hirsutism, microcephaly, heart defects (PDA, VSD), glaucoma, low IQ, speech difficulties, stiff, unsteady gait, hypotonia, attention deficit hyperactivity disorder, down slanting palpebral fissures, maxillary hypoplasia with narrow palate, heavy eyebrows, scoliosis, renal anomalies, hirsutism	Cardiac referral Ophthalmology referral ENT referral Learning and behavioural support Physical therapy
Goldenhar syndrome (Hemifacial microsomia)	Findings usually unilateral. Mandibular hypoplasia, dysplastic or absent ear, pre-auricular tags, hearing problems, macrostomia, epibulbar dermoid. May be vertebral and cardiac defects	Cardiac referral Ophthalmology referral ENT referral OMF/ plastic surgical referral
Cornelia De Lange syndrome	Low birth weight, Synophrys, hirsutism, Beaked philtrum, Heart defects, Limb defects Diaphragmatic hernia	Cardiac referral Ophthalmology referral ENT referral Learning and behavioural support
Pallister Killian syndrome	High birth weight, macrocephaly, diaphragmatic hernia, coarse face, hypotonia, long philtrum, sparse hair over temples, hearing impairment, hypertrophic cardiomyopathy	Cardiac referral ENT referral Learning support
XXY SYNDROME, Klinefelter syndrome	Hypogonadism, Hypogonadism, tall stature, fifth finger clinodactyly, long Legs, low IQ, behavioural problems, autism	Endocrine referral Learning and behavioural support

Achondroplasia	Proximal limb shortening, relatively large head, trident hand, extra skin creases, depressed nasal bridge, lumbar kyphosis Skeletal survey shows square ilia, translucent proximal femur, narrow sacrosciatic notch.	Physical therapy
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7.4 Central hypotonia vs peripheral hypotonia

CENTRAL HYPOTONIA	PERIPHERAL HYPOTONIA
<u>Central nervous system</u> • HIE • Intra cranial hemorrhage • Spinal injury • Infections (congenital/acquired) • Cerebral malformation • Genetic/chromosomal disorders (Trisomy 21, Prader-Willi Syndrome(PWS),Fragile X) • Neuro metabolic disorders :- Leukodystrophies, Lipidoses, Leigh's syndrome • Benign congenital hypotonia	<u>Anterior horn cell</u> • Spinal Muscular Atrophy • Neurogenic arthrogryposis
<u>Metabolic and endocrine</u> • Hypothyroidism • Hypocalcaemia, rickets • Renal tubular acidosis • Organic acidurias • Aminoacidopathies	<u>Neuromuscular junction</u> • Myasthenia gravis (transient/congenital) • Infantile botulism
<u>Connective tissue disorders</u> • Ehlers-Danlos • Marfans syndrome • Osteogenesis imperfecta • Congenital laxity of ligaments	<u>Muscle</u> • Muscular dystrophies (congenital myotonic dystrophy, congenital muscular dystrophy) • Congenital myopathies (central core disease, Nemaline myopathy) • Metabolic myopathies :- Acid maltase deficiency Carnitine deficiency Cytochrome-c-oxidase deficiency Disease of glycogen metabolism
	<u>Peripheral nerves</u> • Hereditary motor and sensory neuropathies • Congenital hypomyelinating neuropathy • Charcot-Marie-Tooth disease • Hereditary sensory and autonomic neuropathy

CLINICAL HISTORY

CENTRAL HYPOTONIA		PERIPHERAL HYPOTONIA
Family History	Consanguinity, Miscarriages, Early child deaths, Family history of development delay, metabolic or genetic disorder	
Antenatal History	Maternal exposure to infections and drugs, IUGR	Polyhydramnios, Reduced fetal movements, Maternal diseases(MG/ Myotonic dystrophy), Family hx of neuromuscular disease
Birth History	Low APGAR, Hx Birth Asphyxia, Birth trauma, Complications at birth – Hypoglycemia, Neonatal seizures, Neonatal Jaundice	Breech presentation/ Malpresentation Prolong ventilation
Past Medical History	Early seizures, abnormal eye movements, apnea or exaggerated irregular breathing patterns.	Normal sleep-wake patterns Recurrent RTI and Respiratory impairment (Myopathy)
Feeding History	Swallowing and feeding difficulties (In infancy with FTT later obesityPWS)	Sucking And Feeding difficulties(SMA, Myopathy)
Development History	GDD Cognitive impairment(DS,PWS)	Selective motor delay with preserved social interactions Development regression

EXAMINATION

Head lag when pulled from supine to sitting position

Full abduction and external rotation of the legs

Flaccid extension of the arms

Older infant W-sitting, a wide-based gait, genu recurvatum, hyper pronation of the feet.

	CENTRAL HYPOTONIA	PERIPHERAL HYPOTONIA
Conscious level	Depressed	Alert
Headshape	Microcephaly	Plagiocephaly
Cry	Irritable /loud cry	Weak cry(Myopathy)
Dysmorphism	Downs syndrome Brachycephaly, flat nasal bridge, up slanting palpebral fissures, small or anomalous ears, short neck with excess nuchal folds, single transverse palmar creases, hypoplasia of the midphalanx of the fifth digit with clinodactyly, joint hyperextensibility Fragile X syndrome Macrocephaly, large ears, epicanthal folds, thickened nasal bridge Praderwilli syndrome narrow bifrontal diameter, strabismus, almond-shaped eyes, enamel hypoplasia, small hands and feet William Syndrome Epicanthal folds, periorbital fullness, stellate irises, and midface hypoplasia Kabuki syndrome	Myopathic face MyotonicDystrophy Facial diplegia, open mouth with tented upper lip, thin cheeks, wasting of temporalis muscles

	long palpebral fissures, eversion of lateral lower eyelid, large protuberant ears, cleft palate, tooth abnormalities, skeletal abnormalities Zellweger Syndrome High fore head, flat face, large anterior fontanel, wide sutures, shallow orbital ridges, broad nasal bridge, epicanthus and micronathia	
Ocular involvement	Cataract, glaucoma (Lowe syndrome)	Extra ocular muscle- sparing (SMA) / weakness-Ptos (MG)
Spontaneous Movements	Paucity of movements	Absent
Supine position	Asymmetry of movements, Fisting of hands, posturing	Frog-legged posture
Ventral suspension		Inverted “U” posture
Vertical suspension	Scissoring	‘Slips through” at the shoulders
Muscle strength	Normal	Reduced
Antigravity movements	Normal antigravity movements	Reduced or absent spontaneous antigravity movements
Weakness	Axial weakness	Profound generalized weakness
Tone	Low	Low
Reflexes	Normal or brisk	Reduced or absent
Clonus	Present	Absent
Fasciculations	Absent	Tongue Fasciculations (SMA)
Primitive Reflexes	Present	Absent
Spine	Scoliosis	Scoliosis
Malformation of other organs	Present	Absent
Muscle wasting	Normal /disuse atrophy	Muscle atrophy
Arthrogryposis	Absent	Present (SMA, Myotonic dystrophy, MG)
Systemic Review CVS	Cardiac murmurs (Downs syndrome, Williams syndrome)	Cardiomyopathy(myotonic dystrophy, metabolic myopathy)
Respiratory		Paradoxical breathing (SMA)
Abdomen	Hepatosplenomegaly (TORCH infection, Lysosomal and Storage disorders) Hepatomegaly (adrenoleukodystrophy) Renal cyst (zellweger syndrome) Undescended testes (Prader -willi syndrome)	
Maternal Examination		Ptos, distal weakness, percussion myotonia and grip myotonia (myotonic dystrophy) Fatiguability of the eyelids with upward gaze or fatiguability of the arms with sustained forward extension (MG)

7.5 Basic Laboratory studies for regressive disorders

Blood

- RFTs, LFTs, ABG, RBS, calcium, phosphate, alkaline phosphatase, uric acid, CPK, lactate, ammonia, and amino acids blood picture

Urine

- Reducing substance, ketone bodies, organic acids

Radiological

- MRI brain, USS Abdomen

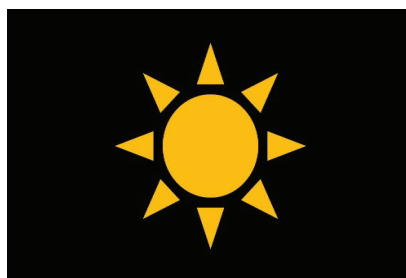
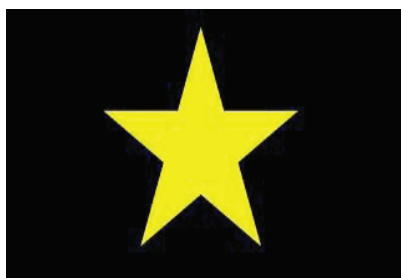
Other:

- ECG, 2D Echo, EEG, EMG
- Hearing and Eye assessment

7.6 NTD Neural Tube Defects

	Thoracic/upperlumbar level (L1)	Mid to high lumbar level (L2-L3)	Lower lumbar (L4-L5)	Sacral level
Mobility	Walking at this level is not common, and mobility is typically at a wheelchair level	Mobility is predominantly using a wheelchair for community distances although those with an L3 level (quadriceps function) have more ability to use a mixed pattern of assisted ambulation and a wheelchair.	Fair to good ambulation potential for both household and community	Ability to ambulate but may need orthoses
Muscle Function	There is no quadriceps function	Hip flexion is present. No gluteus medius/maximus function is present.	Foot dorsiflexion is present.	May involve nerve injury of the cauda equina, so there may be sparing of some distal motor and sensory function
Orthoses	Household/therapeutic ambulation requires the use of a hip-knee-ankle-foot-orthosis (HKFO) or reciprocating gait orthosis	Ambulation requires ankle-foot orthoses (AFO) or knee foot orthoses (KAFO) and usually crutches or a walker	Use of AFOs is common, and an individual may use forearm crutches to improve gait pattern	
Other	Standing may be achieved using passive or dynamic standers		May use a wheelchair for long distances or sports participation	

7.7 CVI friendly picture templates





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