# REHABILITATION OF FLOPPY BABY SYNDROME: OVERVIEW

## Abstract

Floppy baby syndrome is a condition associated with impaired development of motor skills and signs of hypotonia, profound weakness, and rag-doll features. Because numerous disorders can give rise to such symptoms, the diagnosis of the underlying cause of floppy baby syndrome can be complex and problematic. It is critical that all potential causes are examined as early in the course of disease as possible for effective management of these infants. Regardless of whether the underlying cause of hypotonia is peripheral or central in origin, the presentation of floppy infant syndrome focuses on observing for the presence or absence of specific signs such as 'frog-leg' posture, significant head lag on traction or pull-to-sit manoeuvre, or the feeling of 'slipping through the hands' when the infant is held under the arms. Infantile botulism, transient neonatal myasthenia gravis, congenital myasthenia gravis, hypermagnesemia, and aminoglycoside toxicity are all neuromuscular junction disorders that are considered to be a differential diagnosis of floppy infant syndrome. Treatment of the underlying causative syndrome resulting in the presentation of floppy infant syndrome deals with the symptoms of hypotonia, and as a result, the decreased muscle tone, diminished tendon reflexes, any feeding or respiratory difficulties are common. The rehabilitation procedures are very benefits for floppy baby syndrome and this chapter elaborate all the rehabilitation procedure in details.

**Keywords:** Floppy baby syndrome, Hypotonia, Rehabilitation

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## I. INTRODUCTION

The word "Floppy" means decrease in muscle tone; decrease in muscle power and ligamentous laxity and increased range of joint mobility. Floppy infants exhibit poor control of movement, delayed motor skills, and hypotonic motor movement patterns. The child with low tone has muscles that are slow to initiate a muscle contraction, contract very slowly in response to a stimulus. Hypotonia is described as reduced resistance to passive range of motion in joints; weakness is a phenomena in which the maximum power that can be generated is reduced.

Floppy baby syndrome is a rare disorder, thus few epidemiologic studies have been performed to date. The incidence of infantile- and late-onset forms of Floppy baby syndrome has been estimated at 1 in 138,000 births and 1 in 57,000 births, respectively. The overall incidence of this syndrome is estimated at 1 in 40,000. It is transmitted as an autosomal recessive trait and, therefore, affects males and females with equal frequency. In a study of 20 Dutch classic infantile cases and 133 cases reported in the literature, the distribution according to gender was 41% male and 52% female, while the sex was unknown in the remaining 7% of cases, all of which were reported in the literature.

## II. ETIOLOGY

**There are two types of causes namely Central causes and peripheral causes.**

| Central causes | Peripheral causes |
|---|---|
| Cerebral insult such as intracranial haemorrhage | Infantile spinal muscular atrophy |
| Brain malformations | Charcot marie tooth disease |
| Chromosomal abnormalities | Myasthenia gravis |
| Other genetic defects | Congenital myopathy |
| Maternal drug usage | Disorders of glycogen metabolism |

### TYPES

**There are two types of floppy baby.**

| Floppy Strong | Floppy Weak |
|---|---|
| Increased tendon reflex | Hypo or a reflexia |
| Extensor Plantar response | Selective motor delay |
| Sub occipital head growth | Normal head circumference and growth |
| Upper Motor neuron lesion | Lower motor neuron lesion |
| Central hypotonia | Peripheral hypotonia |

## III. PATHOGENESIS

Neuromuscular junction disorders are denoted as causes of peripheral hypotonia that share several features, including hypotonia, facial diplegia, ptosis, feeding difficulties, apnea, respiratory difficulties, generalized weakness, and progressively weakening cry. Each of these neuromuscular junction disorders will be discussed to establish the pathogenesis of

hypotonia and the presentation that leads to floppy infant syndrome. Infantile botulism, caused by consumption of contaminated honey or corn syrup in 20% of the cases, is an age-limited disorder in which Clostridium botulinum (C. botulinum) is ingested, colonizes the intestinal tract, and produces the toxin in situ Infantile botulism usually occurs within six weeks to one year after birth, and the first symptom these infants present with is often constipation. C. botulinum is a gram-positive, spore-forming, obligate anaerobe that is present in the soil worldwide and may spread by dust .The symptoms associated with infantile botulism are all due to the enteric toxins released by C. botulinum. The botulinum toxin is the most potent neurotoxin that does not appear to cross the blood-brain barrier; however, it exerts its toxicity by affecting the transmission at all peripheral cholinergic junctions by interfering with the normal release of acetylcholine from nerve terminals in response to depolarization The enteric toxin causes intestinal immobility and progressive descending paralysis due to the effect on acetylcholine release at the neuromuscular junction and other cholinergic nerve terminals, particularly in the gut Infantile botulism differs from food-borne botulism in the sense that with food-borne botulism, there is ingestion of a preformed toxin in contrast to infantile botulism in which there is continued intra-intestinal production of toxin due to clostridial colonization of the large intestine. Historically, infants afflicted with botulism are between 2 and 26 weeks of age, usually live in a dusty environment adjacent to construction or agricultural soil disruption. As mentioned earlier, while the first symptom of infantile botulism is constipation, other symptoms such as listlessness, ptosis, facial weakness, decreased eye movements, feeding difficulties, and progression to respiratory failure can occur.
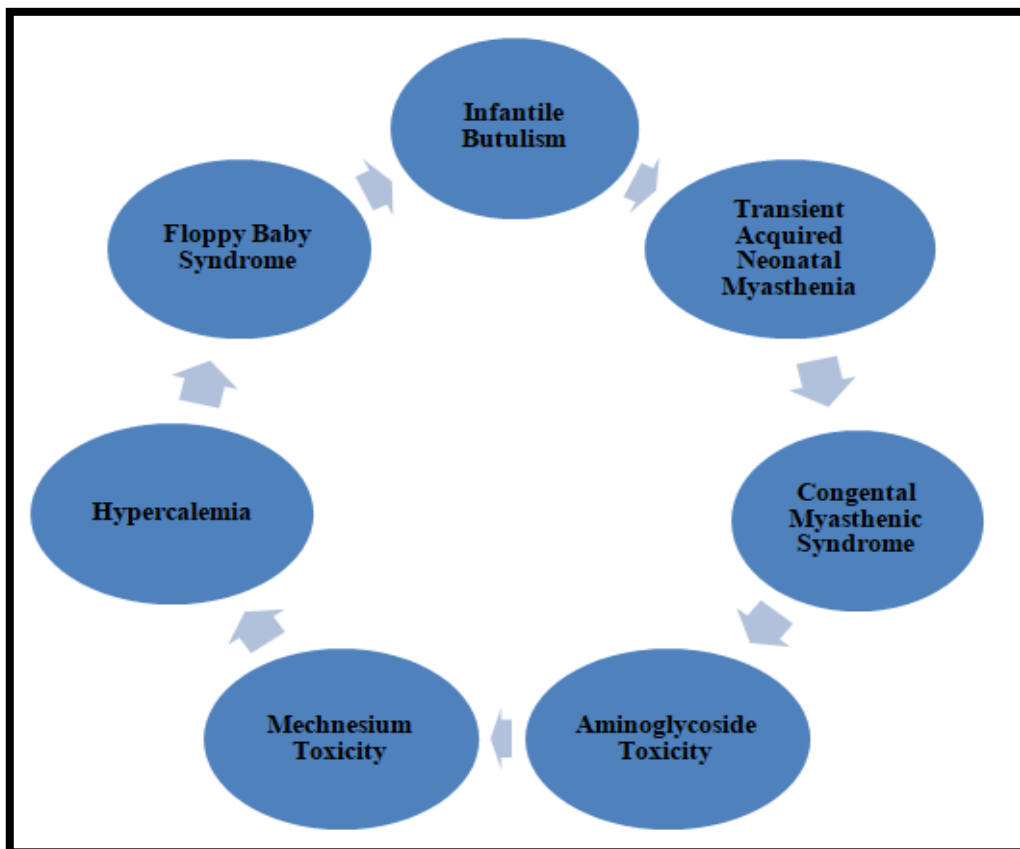


**Figure 1: Pathophysiology of Floppy Baby Syndrome**

Congenital myasthenic syndromes result from gene mutations affecting the neuromuscular junction structure and function. Infants presenting with the myasthenia syndrome share several features, including hypotonia, facial diplegia, ptosis, feeding difficulties, apnea, respiratory difficulties, generalized weakness, and a progressively weakening cry, making congenital myasthenia syndrome a differential diagnosis of floppy infant syndrome .Congenital myasthenic syndromes can present at any time from birth to adulthood, though usually within the first two years of life, and result in a spectrum of diseases ranging from mild weakness to severe disability with life-threatening episodes .Congenital myasthenia is an umbrella term for a category of syndromes that all impact the neuromuscular junction but differ in whether the deficiency is due to presynaptic, synaptic, or postsynaptic defects of neuromuscular transmission which leads to either an increased response to acetylcholine or a decreased response. Gene changes altering the structure and function of the neuromuscular junction cause congenital myasthenic disorders. Congenital myasthenia syndrome is a differential diagnosis for floppy infant syndrome because infants with the disease share a number of characteristics, including hypotonia, facial diplegia, ptosis, feeding issues, apnea, respiratory issues, generalised weakness, and a progressively fading scream. Congenital myasthenic syndromes can cause a spectrum of illnesses, from minor weakness to severe impairment and life-threatening crises, and can manifest at any time from birth to adulthood, though typically within the first two years of life. The phrase "congenital myasthenia" refers to a group of diseases that all have an effect on the neuromuscular junction but differ depending on whether there is a deficit in presynaptic, synaptic, or postsynaptic neuromuscular transmission.

Transient acquired neonatal myasthenia occurs in infants born to mothers with myasthenia gravis in which the acetylcholine receptor antibody that causes myasthenia gravis crosses the placenta and exerts a blocking effect that is responsible for the interference with neuromuscular transmission. Neonatal transient myasthenia gravis is a self-limited disorder that may be potentially life-threatening if prompt and accurate diagnosis and supportive respiratory management are not initiated. There is a natural passive transfer of maternal antibodies that cross the placenta and bind to fetal motor-end plates, specifically against the nicotinic acetylcholine receptor (AChR).Transient neonatal myasthenia was reported in 12.26% of infants born to mothers with generalized myasthenia gravis before the discovery and use of AChR antibody titers for diagnosis of acquired autoimmune myasthenia gravis.

Aminoglycosides are a mainstay of antimicrobial therapy for infants in cases in which infections are due to gram-negative bacteria, accounting for up to 25% of all sepsis episodes in neonatal units. Aminoglycosides have a narrow therapeutic window, and close monitoring is required to minimize potential nephrotoxicity, ototoxicity, neuromuscular blockade. Out of the aminoglycosides, gentamicin, neomycin, streptomycin, tobramycin, and kanamycin have been reported to produce clinically significant muscle weakness on occasion in non-myasthenia gravis patients. Aminoglycoside toxicity is a concern of greater magnitude in premature infants and neonates, and this is mainly due to the renal system being immature and thus, resulting in a prolonged serum half-life of the aminoglycosides. The prolonged serum half-life will manifest as nephrotoxicity, ototoxicity, and also cause neuromuscular blockade resulting in muscle weakness, generalized hypotonia ultimately making aminoglycoside toxicity a differential diagnosis of floppy infant syndrome.

Elevated magnesium levels can be encountered in the new born following the treatment of maternal eclampsia with magnesium sulphate or following the use of magnesium antacids in the new born, resulting in an encephalopathic infant with hypotonia, depressed deep tendon reflexes, abdominal distension due to ileus and irregularities of cardiac rhythm . Hypermagnesemia is defined as a serum magnesium concentration greater than 1.15 mmol/L (2.8 mg/dL) . Women with pre-eclampsia are at risk of developing seizures, which are associated with adverse outcomes for the mother and the fetus, and therefore, anti-convulsant treatments such as magnesium sulphate are given to mothers with eclampsia to reduce the risk of seizures and improve outcome. Infants born to mothers with pre-eclampsia or eclampsia who received magnesium sulphate can have hyper magnesemia presenting with generalized hypotonia, apnea, bradycardia, feeding difficulty, and, in severe cases, respiratory distress and may even mimic septic shock. The combination of symptoms that the infants present with could be described as floppy infant syndrome. The feeding difficulty and respiratory distress could be allocated to the generalized hypotonia causing decreased muscle tone and, thus, decreased muscular movements. Magnesium is known to inactivate acetylcholine at the neuromuscular junction, especially in the respiratory muscles, and does not affect the brain directly.

Hyperkalemia is present in up to 52% of premature infants with a birth weight of less than 1000g, and hyperkalemic infants are at a high risk of developing life-threatening cardiac arrhythmias . Non-oliguric hyperkalemia is characterized  by an excessive increase in serum potassium concentration at 24 hours after birth and is mainly due to the immature functioning of the sodium ($Na^+$)/potassium ($K^+$) pump . The early-onset hyperkalemia may have been caused by the accumulation of potassium ions transported through the placenta, the shift of potassium ions from the intracellular to the extracellular space in the infant due to the malfunctioning of the $Na^+/K^+$ pump and the inhibition of renal distal tube potassium ion secretion . The causes of hyperkalemia in infancy include acute hemolysis, kidney disorders, and hormonal disorders.

## IV. SIGNS AND SYMPTOMS

1. Abnormal posturing of limb and body
2. Diminished resistance of limb to passive movements.
3. Abnormal joint range of motion
4. Delayed motor milestones
5. Paradoxical breathing pattern
6. Frog like posture
7. Weakness in the anti-gravity muscles
8. Poor swallowing ability
9. Inability to cough
10. Myopathic faces.
11. Decreased muscle tone; muscles feel soft and doughy
12. Ability to extend limb beyond its normal limit
13. Failure to acquire motor-related developmental milestones (such as holding head up without support from parent, rolling over, sitting up without support, walking)
14. Problems with feeding (inability to suck or chew for prolonged periods)
15. Shallow breathing
16. Mouth hangs open with tongue protruding (under-active gag reflex)

17. Central nervous system function and intelligence in children is normal.
18. Some children acquire gross motor skills (sitting, walking, running, jumping) more slowly than most.
19. Ptosis and external ophthalmoplegia in a floppy weak child suggestive of myasthenia gravis
20. Paradoxical breathing pattern. Intercostal muscles paralyzed with intact diaphragm.
21. Inverted U position
22. The back hangs over the examiner's hand, and the limbs and head hang loosely
23. Passive hyper extension of the legs
24. Pull to sit-Head lag
25. Decreased tone of the shoulder girdle allows the infant to slip through the examiner's hands

## V. CLINICAL PRESENTATIONS:

On a routine physical exam of an infant with subtle signs of developmental delay, an astute paediatrician may hear a heart murmur or gallop rhythm, which can lead to further work-up and diagnosis. Parents also often report easy fatigue and sweating during feeding. On occasion, a child may present with a supraventricular tachycardia and other arrhythmic disturbances. On the basis of clinical experience, infants with Floppy Baby syndrome may present for pulmonary, neurological, gastrointestinal, or cardiac issues. In the classic pulmonary presentation, there are frequent upper respiratory tract infections which are treated with antibiotics. When a satisfactory treatment response is not obtained, a chest x-ray is often ordered to look for underlying pneumonia showing massive cardiomegaly. Alternatively, a child may present with respiratory insufficiency due to repeated infections. $CO_2$ retention may be found when blood gases or clinical chemistries are analyzed, prompting further evaluation.
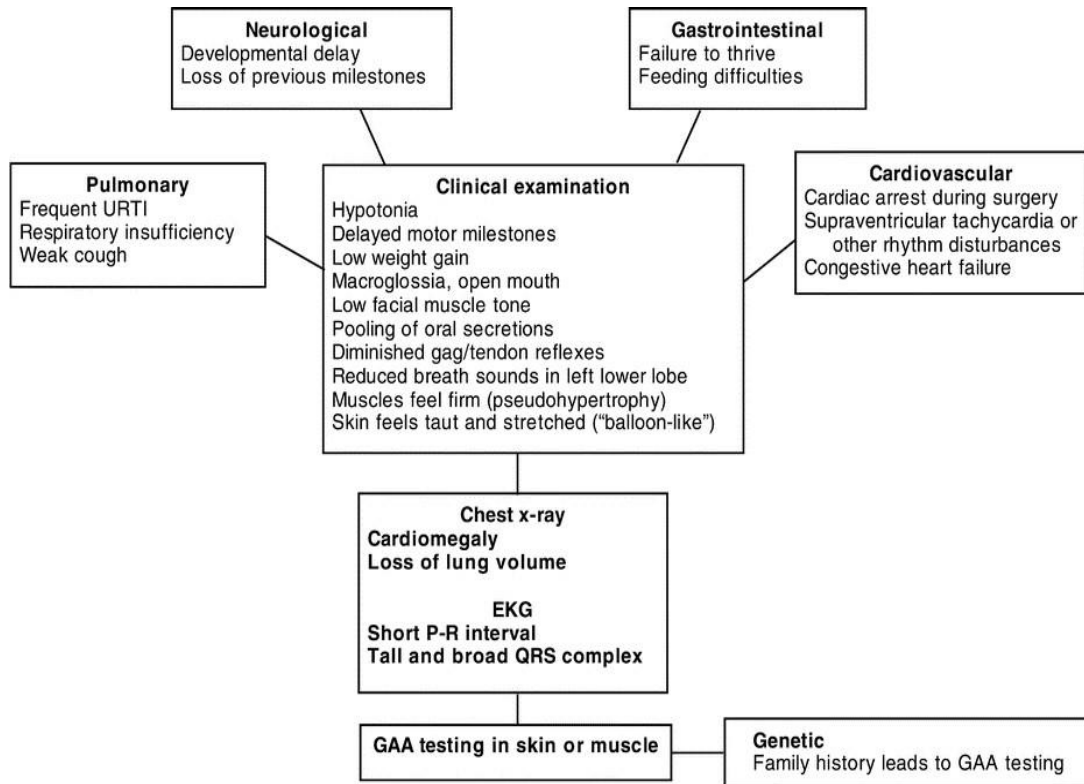
In a neurological presentation, parents bring their child to the paediatrician with concerns regarding development typically at about 3–4 months of age. The child may be referred to a paediatric neurologist due to concerns of severe hypotonia and developmental delay. In some cases, a diagnosis of mitochondrial disease is considered, because of the multisystemic nature of organ involvement (cardiomegaly, hepatomegaly, hypotonia) in which these disorders may present. Alternatively, an infant may come to clinical attention following loss of previously achieved milestones.

## 1. Examination

- Palpation of the muscles will be flabby
- Adductor angle : Angle between thighs when hips maximally abducted with extension at knees
- Popliteal angle: Hips flexed on the abdomen but holding of the knees
- Scarf sign: Flexed at the elbow pull across the chest but holding at the hand and wrist.
- Heel to ear Manoeuvre: both extended legs lifted towards the ears without lifting the pelvis.

## 2. Diagnosis

- If central hypotonia is suspected: MRI, CT scan, karyotyping, Molecular genetics
- If peripheral hypotonia is suspected: EMG, Nerve conduction study, Muscle biopsy, CK level.

**Neurological**
Developmental delay
Loss of previous milestones

**Gastrointestinal**
Failure to thrive
Feeding difficulties

**Pulmonary**
Frequent URTI
Respiratory insufficiency
Weak cough

**Clinical examination**
Hypotonia
Delayed motor milestones
Low weight gain
Macroglossia, open mouth
Low facial muscle tone
Pooling of oral secretions
Diminished gag/tendon reflexes
Reduced breath sounds in left lower lobe
Muscles feel firm (pseudohypertrophy)
Skin feels taut and stretched ("balloon-like")

**Cardiovascular**
Cardiac arrest during surgery
Supraventricular tachycardia or
  other rhythm disturbances
Congestive heart failure

**Chest x-ray**
**Cardiomegaly**
**Loss of lung volume**

**EKG**
**Short P-R interval**
**Tall and broad QRS complex**

**GAA testing in skin or muscle**

**Genetic**
Family history leads to GAA testing

## 3. Management

- It is very important to continue supportive care with regards to feeding and respiration. Most of these hypotonic neonates need prolonged mechanical ventilation. Regular physiotherapy is needed to aid the clearance of respiratory secretions and will prevent limb contractures. It is vital to aggressively treat any respiratory infections. Feeding should be initiated by nasogastric tube and gastrostomy may be needed for few babies. Weight should be closely monitored as excessive weight gain can worsen existing muscle weakness.

- Children with neuromuscular disorders need attention if they require anesthesia. Muscle relaxants should only be used if necessary as they have a more prolonged effect in these children. They are also susceptible to malignant hyperthermia and implicating agents should be avoided.

- Orthopaedic intervention in setting of established or evolving joint contractures.

- Feeding: Nasogastric feeding, caloric supplementation, gastrostomy. Low-tone infants often have difficulty feeding, especially coordinating the suck-swallow reflex required

for proper breastfeeding. Early diagnosis of hypotonic newborns can help mothers find the support and information they need to establish a successful breastfeeding relationship. Hypotonic babies may take longer to breastfeed because of the poor timing of sucking bursts and the need for long rests. If feeding is inefficient, they will also require greater feeding frequency. A baby with low muscle tone may suck better when the head and bottom are level, indicating pillow support in the lap. If the infant tends to arch his back, it may be helpful to swaddle the child loosely with arms drawn across the chest and legs drawn up toward the belly with a rounded spine during feedings.

- It is important to ensure multidisciplinary follow-up for neonates with neuromuscular disorders. Follow-up should be arranged with neurologist and respiratory team, and an appointment with the geneticist for genetic counselling should be offered.

- Psychological approach to encourage of overall development and stimulation of learning and counselling to the parents.

- Position to encourage with help of physiotherapist to carrying techniques and head control procedure should maintained properly.

- Physiotherapy: Regular physiotherapy will prevent contractures. Physical therapists might use neuromuscular/sensory stimulation techniques such as quick stretch, resistance, joint approximation, and tapping to increase tone by facilitating or enhancing muscle contraction in patients with hypotonia.

- Physiotherapy for floppy infants is aimed at stimulating normal movement, co-ordination and strength through use of play and functional activities. This makes it more fun for children, more meaningful in terms of everyday activity, and reproducible at home and school. It should encourage as much involvement from parents and carers as possible in their child's rehabilitation, in order for them to feel very much part of the process. It should be able to give parents specific activities that will help to develop their child's skills and movement control, and tailor rehabilitation to the needs of individual children.

- Some physiotherapy techniques also involve a very hands-on approach in order to change soft tissue length and tension, and facilitate normal movement patterns. Specific stretches, massage techniques and correct handling can be taught to parents in order that the treatment outcome is the most effective it can be. Biofeedback, carrying position, balance and gait training, hydrotherapy, therapeutic horseback riding, rebound therapy.

- Occupational therapy teaches the skills needed to carry out day-to-day activities. For example, the occupational therapist may focus on improving the hand and finger skills needed for dressing and feeding.

- A speech and language therapist can assess child's feeding and swallowing, and help identify swallowing problems (dysphasia) that can sometimes be associated with hypotonia.

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