



Case Learning: The floppy infant

<p>Situation</p>	<p>A baby had poor weight gain in the first few months of life. They were reviewed several times for this and provided with feeding support. When they were 6 months old, there were concerns about limited motor development, and they were referred to Paediatrics. At 7 months of age, they were diagnosed with Type 1 Spinal Muscular Atrophy (SMA). Supportive care was provided, and they later sadly died before their second birthday. Type 1 SMA is a rare, monogenic, progressive neuromuscular condition. Without any treatment, children will not survive infancy, or will need permanent ventilatory support by 2 years of age. Treatments that target the altered genes causing SMA are now available, substantially improving event-free survival and motor development, however early lower motor neuron damage is irreversible.</p>
<p>Takeaway</p>	<p>Early identification of hypotonia or delayed motor development, urgent referral & rapid diagnosis is vital to halt the progress of SMA Type 1 and provide affected children with optimal outcomes.</p>
<p>Actions</p>	<div style="display: flex; flex-direction: row;"> <div style="flex: 1;">    </div> <div style="flex: 2;"> <ol style="list-style-type: none"> 1. Pick up the baby <ul style="list-style-type: none"> • Head lag, hypotonia and reduced antigravity movements can only be observed when a baby is lifted up. • Babies with SMA Type 1 are often socially responsive and alert, which can provide false reassurance about their development. • When seeing a child under 6 months of age, ask the parent/carer to pick up the baby out of their car seat/infant carrier/pram. 2. Pick up the monitoring <ul style="list-style-type: none"> • Ask the Health Visitor to monitor development if there are any concerns that do not warrant immediate action. 3. Pick up the phone <ul style="list-style-type: none"> • Any floppy infant (signs of hypotonia including head lag, limited antigravity movements, floppiness evident on being held prone, weak cough/cry, feeding difficulties) or if you are concerned about possible SMA: <ul style="list-style-type: none"> ➤ Immediate same day referral to the on-call General Paediatrician. </div> </div>
<p>Resources</p>	<p>Institute for Health Visiting Good Practice Points: Identifying, referring & supporting infants with Spinal Muscular Atrophy Type 1.</p> <p>Sheffield Childrens Hospital SMA Gene Therapy Service</p> <p>Spinal Muscular Atrophy UK</p>

For more information about the LLR Child Death Overview Panel [click here.](#)