Aim
To provide consistent management and follow up to infants and families of infants with Achondroplasia.

Background
Achondroplasia is the most common cause of disproportionate short stature, with 3-5 children born with the condition each year in WA. It is characterised by rhizomelic short stature, macrocephaly, frontal bossing and midface retrusion. Life span is normal, although craniocervical junction compression increases the risk of death in infancy.

It is inherited in an autosomal dominant manner with complete penetrance. Around 80% of individuals have achondroplasia as a result of a new mutation (de novo) and have parents with normal stature.

It is not easily identified on routine ultrasound during pregnancy, therefore it is most commonly diagnosed either later in pregnancy when a scan is done for another reason, or at some point after the baby is born.

The diagnosis of Achondroplasia can be established solely based on the characteristic clinical and radiological features. FGFR3 gene testing is done in uncertain cases, for confirmation of the clinical diagnosis and in prenatal settings when achondroplasia is suspected.

Risk
Infants with Achondroplasia may not receive the correct management and follow-up.

Clinical Description
The figure below represents the common clinical features and medical complications in individuals with Achondroplasia.
Radiological Features

1. Short and robust tubular bones.
2. Proximal femoral radiolucency.
3. Narrowing of the interpedicular distance of the lumbosacral spine (normally widens in a caudal direction).
4. Square ilia and horizontal acetabulae.
5. Narrow sacrosciatic notch.
6. Mild generalised metaphyseal changes.
7. Brachydactyly / trident configuration of fingers.

Antenatal Consultation
If the diagnosis of achondroplasia is suspected or known antenatally the mother should be offered an antenatal consultation with a Neonatal Consultant to develop a Neonatal Management plan for the baby.

Neonatal Management

- Maintain appropriate handling of baby, by parents and staff to protect neck/airway:
  - Cot card highlighting the need for careful handling.
  - Positioning and handling of babies with Achondroplasia after birth
      (to be printed and placed in plastic sleeve on cot).
Achondroplasia

- Measure and plot weight, length and head circumference on Achondroplasia-specific growth charts. 
- Clinical examination by Senior Neonatal Registrar or Consultant in the first 24 hours.
  - Hypotonia, apnoea, feeding, colour change.
- Request for postnatal MRI cervical spine / craniocervical junction /brain (feed and wrap) – prior to D/C if feasible or within first 6 weeks.
- Request Genetic review as inpatient.
- Skeletal survey (if diagnosis suspected but not yet confirmed - discuss with Genetics if diagnosis is unclear).

Parental Education Prior to Discharge

- Physiotherapist / ward staff on site – handling guidelines / demonstration.
  - Paediatric Physio page 83121 (weekdays), e-referral to KEMH Physio (Tick Urgent – attention Natasha Amery)
- CPR education / safe sleep guideline.
- Rural / regional families – advice re having an adult travelling in back seat with baby during trip home and suitable breaks if journey a significant distance.
- Car seat assessment by physiotherapist before discharge.

Discharge Planning

Referrals to be made by Neonatal Team

1. PCH Rehabilitation Medicine Early Intervention Service (Achondroplasia) (state "Diagnosis Achondroplasia" in referral).
2. Genetic Services via e-referral from public hospitals or through the Central Referral Service (state “Diagnosis Achondroplasia” in referral)
3. PCH Sleep Service (state “newborn with achondroplasia, for sleep study at 6 weeks of age”). If more urgent (Respiratory concerns, abnormal craniocervical junction MRI) – discuss with Respiratory consultant on call.

Hearing assessment – refer to PCH Audiology/ENT if failed newborn screen.

Provide information about available resources

i. https://sspa.org.au/ (Parent representative for WA: Suzann Franklin landsfranklin@bigpond.com; 08 90413220)
ii. https://medlineplus.gov/genetics/condition/achondroplasia/

References and related external legislation, policies, and guidelines


Useful resources (including related forms)

- Consent to Genetic Testing
- Consumer information on Genetic Testing
  - https://sspa.org.au/ (Parent representative for WA: Suzann Franklin landsfranklin@bigpond.com; 08 90413220)
  - https://medlineplus.gov/genetics/condition/achondroplasia/
This document can be made available in alternative formats on request for a person with a disability.

<table>
<thead>
<tr>
<th>Document Owner:</th>
<th>Neonatology</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reviewer / Team:</td>
<td>Neonatology / Genetics / Respiratory Medicine / PCH Rehabilitation</td>
</tr>
<tr>
<td>Date First Issued:</td>
<td>February 2021</td>
</tr>
<tr>
<td>Last Reviewed:</td>
<td>10th March 2021</td>
</tr>
<tr>
<td>Amendment Dates:</td>
<td></td>
</tr>
<tr>
<td>Next Review Date:</td>
<td>10th March 2024</td>
</tr>
<tr>
<td>Approved by:</td>
<td>Neonatal Coordinating Group</td>
</tr>
<tr>
<td>Date:</td>
<td>23rd March 2021</td>
</tr>
<tr>
<td>Endorsed by:</td>
<td>Neonatal Coordinating Group</td>
</tr>
<tr>
<td>Date:</td>
<td></td>
</tr>
<tr>
<td>Standards Applicable:</td>
<td>NSQHS Standards: 📋📊🔍🔍</td>
</tr>
<tr>
<td></td>
<td>Child Safety Standards 1,10</td>
</tr>
</tbody>
</table>

Printed or personally saved electronic copies of this document are considered uncontrolled.