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PIERRE ROBIN SEQUENCE IN THE UK & ROI

(Short Study Name: Pierre Robin Sequence)

Abstract

Pierre Robin Sequence (PRS) is a congenital condition characterised by cleft palate, micrognathia, and glossoptosis. Together, these abnormalities can lead to upper airway obstruction of variable severity which is typically evident in the neonatal period but can be subclinical or delayed in onset. If not recognised and optimally managed, upper airway obstruction can lead to growth faltering and respiratory failure.

Limited data is available about the current incidence of PRS; no epidemiological studies have been published in the UK for over 30 years, and birth prevalence reports from other countries vary widely from 1 case per 3000 to 14 000 live births.

The management of airway obstruction and growth faltering in children with PRS appears to vary significantly between treatment centres, with no evidence-based treatment guidelines currently in place to inform best practice.

This study aims to provide an updated estimate of PRS birth prevalence in the UK and Republic of Ireland (ROI), and to ascertain whether there are geographical variations in its incidence. We will also describe the current management practices in use in the UK and clinical outcomes at 1-year follow-up.

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Website

www.rcpch.ac.uk/bpsu/PRS

Background

PRS is a condition present from birth with three main features: a small lower jaw (micrognathia), backward positioned tongue (glossoptosis), and defect of the roof of the mouth (cleft palate). Together, these abnormalities cause a narrowing of the pharyngeal space, which may lead to upper airway obstruction. Typically, this causes breathing and feeding difficulties in the neonatal period and early childhood. Mild cases can be managed by nursing the infant in a position that best opens the airway, and by using specialised feeding bottles. More severe cases may need temporary feeding tube or artificial airway placement, whilst those worst affected require surgical procedures to maintain a safe airway until 'catch-up growth' of the mandible occurs.

The aetiology of PRS is not fully understood, but is likely to be multifactorial including genetic factors and exposure to toxins or compressive forces in-utero.

PRS diagnosis has far-reaching consequences for affected children, their families, and responsible healthcare teams. Infants often spend many weeks in hospital and need long-term support from a large multidisciplinary team. Failure to optimally manage breathing and feeding problems can cause respiratory failure and poor growth. Long-standing airway obstruction can result in impaired oxygen delivery, leading to right heart failure and neurodevelopmental problems.

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There is currently little research evidence available regarding the extent of the problem posed by PRS, or how best to manage it. Data regarding its incidence are limited, and no UK epidemiological studies of PRS have been published for over 30 years. Management is currently variable between treatment centres, with no evidence-based guidelines in place to standardise clinical practice.

Coverage United Kingdom and the Republic of Ireland

Duration January 2016 to January 2017 (13 months of surveillance) with a 12 month follow-up.

Research Questions

- To identify the current birth prevalence of PRS in the UK and ROI, including identification of any geographical variations.
- To outline the clinical presentation, initial management, and 12 month clinical outcomes of children with PRS in the UK and ROI.
- To describe the current airway and feeding management practices in hospitals across the UK and ROI, including variations in practice between treatment centres.
- To compare clinical outcomes between the various airway management modalities available, using the following outcome measures where available; duration of initial hospital admission, growth, 12 month neurodevelopmental outcomes, and sleep study findings.

Case definition

Any live-born infant, born in the UK or ROI during the surveillance period (January 2016 - January 2017), with the following clinical features:

- Cleft palate

AND

- Micrognathia/ retrognathia or glossoptosis

AND

- Evidence of resulting compromise, with <u>at least one</u> of the following features:
  - i) Signs of upper airway obstruction
  - ii) Feeding difficulties
  - iii) Faltering growth (loss of >10% birth weight in the first week of life, or fall across two centile lines on a standardised growth chart)

Reporting instructions Please report any child meeting the above surveillance case definition who you have seen for the first time during the last month.

Methods Each paediatrician reporting a child who meets the above case definition of PRS will be sent a clinical questionnaire by the study team, which can be returned either by post or email. A second questionnaire will be sent 12 months later to collect follow-up data.

Throughout the study, all patient data will be dealt with in strict confidence, and affected children and their families will not be contacted directly by the PRS study team at any stage.

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Ethics approval This study has been approved by NRES South East Scotland Research Ethics Committee 2 (REC reference: 15/SS/0049; IRAS project ID: 161997) and has been granted Section 251 HRA-CAG permission (CAG Reference: 15/CAG/0141)

Support group Cleft Lip and Palate Association

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