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What is 'Pierre Robin sequence'?☆

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Summary Different nosology has hampered our understanding of patients with Robin sequence. Defining this disorder correctly has important consequences for physicians and parents. While reviewing treatment options for Robin sequence we were surprised to see how often different definitions were used to describe this condition. This prompted us to perform a review into the current understanding when diagnosing and defining this disorder. At our Annual Dutch Cleft Palate Meeting a questionnaire was given to all those attending requesting them to summarise characteristics needed for a definition of 'Robin sequence'. Sixty-six questionnaires were returned, demonstrating 29 different descriptions. Our study demonstrates that there is widespread confusion in the Netherlands defining Robin sequence. This lack of uniformity in the definition hampers the comparison of outcome studies. The treatment of patients with Robin sequence often involves multidisciplinary involvement, making it crucial to have one common definition. We suggest using the description originally summarised by Pierre Robin consisting of micrognathia, glossoptosis and airway compromise. Once the diagnosis of Robin sequence is made other adjuncts such as an associated cleft palate, syndrome or feeding problems could be added to the description.

> 'As these children generally present vagosympathico-endocrine instability, they walk and talk late. They are often considered lazy, when they are in reality merely sick children who will become normal and active after proper treatment'
>
> (Pierre Robin, 1934)

In 1923 the Parisian stomatologist, Pierre Robin, described a range of findings consisting of breathing problems in patients with glossoptosis and associated micrognathia.[1] Only in a later manuscript did Robin mention that patients with the described findings could have an associated cleft palate.[2] Following the descriptions made by Robin, several other clinicians added valuable information and since the 1960s this condition has been known as Pierre Robin syndrome.[3–5] Although this well-known eponym has had several name changes, this condition is often described as being Pierre Robin sequence, syndrome, complex or anomaly. The incidence of this condition is uncertain but it

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is estimated to occur in 1 in 8500 to 1 in 20 000 births.[4–6] Even today this condition is still associated with significant mortality.[4–7] Specificity in the diagnosis is essential for correct advise about prognosis (e.g. mandibular outgrowth)[8] and treatment modalities. While reviewing treatment options for Robin sequence we were surprised to see how often different definitions were used to describe this condition. This prompted us to perform a review into the current understanding when diagnosing and defining this disorder.

Patient and methods

At the annual meeting of the Dutch Cleft Palate and Craniofacial Association held in Utrecht, the Netherlands (November, 2006), all attending members of the cleft palate teams were given a questionnaire about Robin sequence. The questionnaire evaluated the precise characteristics needed for diagnosing 'Pierre Robin sequence'. In the questionnaire open questions were asked without examples to choice from, and all those attending where given a free choice about the possibilities.

Results

Sixty-six questionnaires where returned and 29 different descriptions were found. Table 1 reveals the 10 most common descriptions. The most common range of findings necessary for diagnosing Robin sequence where retrognathia (RG) and cleft palate (CP) ($n = 10$), RG, CP and breathing problems ($n = 6$), micrognathia (MG) and CP ($n = 6$), MG, CP and breathing problems ($n = 5$) and RG, CP and glossoptosis ($n = 5$). Other descriptions included: MG/CP/cardiac + chromosomal abnormalities; RG/CP/ microstomia; MG/CP/ pharyngeal hypotonia; MG/macroglossia, etc. Table 2 demonstrates that, of the 18 plastic surgeons attending, 12 different characteristics were described. Ten different findings where described by the speech pathologists attending. The seven maxillofacial surgeons that attended all had different definitions. The other different findings described per discipline are summarised in Table 2. Table 3 demonstrates that most did not think the difference between micrognathia and retrognathia makes a difference in defining this disorder.

Table 1 Overview of the 10 most common descriptions seen in the questionnaire.

Diagnosis	*n*	% of total
1. RG/CP	10	15
2. RG/CP/breathing problems	6	9
3. MG/CP	6	9
4. MG/CP/breathing problems	5	7.5
5. RG/CP/GT	5	7.5
6. MG/GT/CP	4	6.0
7. CP/MG/macroglossia	3	4.5
8. MG/CP/abnormal tongue implant	3	4.5
9. MG	2	3
10. RG/CP/frenulum	2	3

Table 2 Different findings per discipline.

Discipline	Attending ($n = 66$)	Different findings described
Plastic surgery	18	12
Speech pathology	14	10
Orthodontics	11	7
Maxillofacial surgery	7	7
ENT	5	5
Psychology	4	3
Paediatrics	3	3
Embryology	1	1
Dentist	1	1
Medical student	1	1
Unknown	1	1

Discussion

This study demonstrates that in the Netherlands there is no uniformity among physicians involved in cleft care in defining this well-known eponym. Without a common definition of the disorder it will be difficult to compare different treatment options.

Despite the fact that several other authors described this condition before Pierre Robin, clinicians have honoured the contributions Robin made to the literature by establishing the eponym 'Pierre Robin syndrome'.[3] However, in medicine it is unusual to use the first name of a person after whom a medical condition has been named. We subsequently agree with the purists and suggest that we call this condition 'Robin sequence' and not 'Pierre Robin sequence'.

A patient with a syndrome is defined as one who has multiple anomalies with all of those anomalies having a single pathogenesis.[9] A patient that has a sequence also has multiple anomalies, but all or some of the anomalies are caused secondarily by one of the anomalies present in that person.[5] It is generally agreed that primary pathology is the micrognathia. Because of abnormal mandibular outgrowth (micrognathia), the tongue stays high and retroposed and impinges against the nasopahrynx causing breathing problems and impeding feeding. During normal palatal development, the tongue lies between the two

Table 3 Overview of answers to question: ''Will difference between 'micrognathia' and 'retrognathia' influence your decision making process?''.

	No	Yes
Plastic surgery	16	8
Maxillofacial	5	1
ENT	8	1
Speech	6	1
Orthodontics[a]	2	4
Paediatrician[a]	4	1
Nurse	8	3
Genetics	1	1
Unknown	0	1

[a] One respondent did not respond to this question.