

## Congenital Retrognathia and the Pierre Robin Sequence

Peter M. Dunn, MA, MD, FRCP, FRCOG, FRCPCH  
Emeritus professor of perinatal medicine  
and child health, University of Bristol, UK  
e-mail: P.M.Dunn@bristol.ac.uk

Congenital retrognathia, giving the newborn infant the appearance of having no chin, is caused by sustained prenatal acute flexion of the head onto the anterior upper chest. This leads to retarded forward growth of the fetal mandibles. Sometimes there is evidence of reciprocal pressure on the chest. Exceptionally there may even be a deep depression on the upper sternum (see Figure 1 below).

The presence of retrognathia during prenatal life causes the fetal tongue to be



Fig 2a

Baby W at birth (with parental permission) Head flexed onto chest.

accommodated backwards and upwards into the nasopharynx. This may lead to separation of the palatal folds and a midline cleft palate (Fig 2).

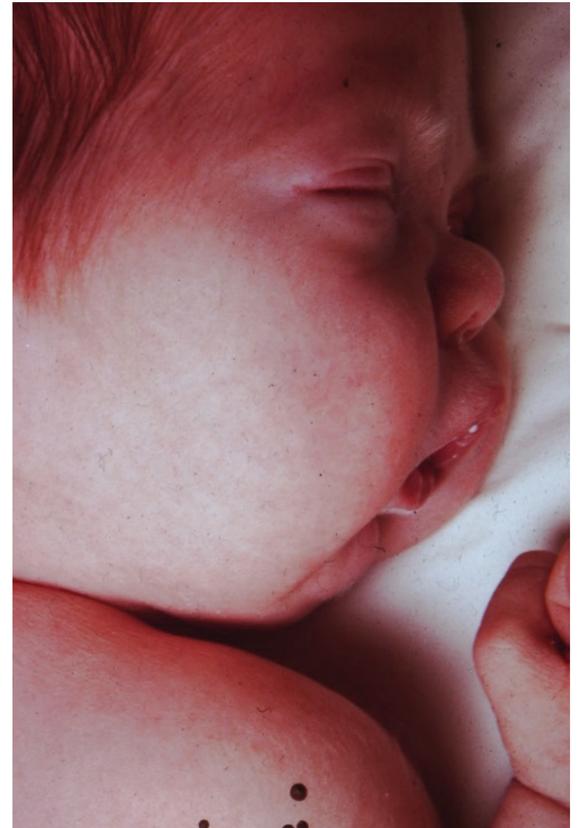


Fig 2 b

Baby W at birth (with parental permission) Showing retrognathia



Figure 1a

Baby M at birth (with parental permission) Head flexed onto upper chest wall.



Fig 2 c

Baby W at birth (with parental permission) Showing midline cleft palate



Figure 1b

Baby M at birth (with parental permission) Head deflexed, showing deep cavity over upper sternum.



Fig 2d

Baby W (with parental permission) Aged 18 months, following repair of palate at 6 months. The retrognathia is much less apparent.

## Congenital Retrognathia and the Pierre Robin Sequence



Fig 3

Baby with Trisomy 18. Died day 1.  
Midline cleft palate secondary to retrognathia giving appearance of pressure disruption.



Fig 5

Baby nursed in the prone position with head suspension due to Pierre Robin glossoptosis.

In other cases, the palate may give the impression of having broken down due to pressure after previous fusion (Fig 3). Alternatively, the palate may just be high arched. Following birth the baby may exhibit respiratory tract obstruction due to glossoptosis<sup>(1,2)</sup>.

The combination of congenital retrognathia, midline cleft palate and glossoptosis was first described by Pierre Robin in 1926. He incorrectly attributed the mandibular hypoplasia to syphilis or to tuberculosis and it was not until 1933 that Davis and Dunn<sup>(3)</sup> suggested that it was the result of sustained flexion of the fetal head in-utero. The combination was first called the Pierre Robin Syndrome (Fig 4). More recently it has been renamed the Pierre Robin Sequence<sup>(4)</sup>. Management is mainly directed towards securing an airway and assuring an adequate

milk intake. Prone nursing with or without suspension of the head usually proves adequate (Fig 5). Over time there is catch-up growth of the mandibles (Fig 2d).

Over a period of 30 years the author has looked after eleven cases of Pierre Robin Sequence, suggesting an incidence of around 1 case/10,000 births. This is similar to the incidence reported by Maas and Poets (2014)<sup>(5)</sup>.

The belief that the Pierre Robin Sequence was due to pressure constraint in-utero, received powerful support from the work of Poswillo (1968)<sup>(6)</sup>. He showed that early amniotic sac rupture of rat embryos at 15½ days gestation (equivalent to seven weeks gestation in the human pregnancy) led to retrognathia (97%) and midline cleft palate (100%) as well as the presence of other moulding deformities. Among the author's eleven cases of the

Pierre Robin Sequence, seven exhibited evidence of congenital postural deformation, including congenital dislocation of the hip (in 2 cases), congenital postural scoliosis (2), deformity of the hand (1), deformity of the chest (1), and congenital talipes (5). The pregnancies of these babies often revealed the typical characteristics that are associated with deformation such as primiparity and oligohydramnios. Two of the series were also malformed; one a case of trisomy 18 and one with congenital heart disease. Both died. Eight of the remaining nine infants survived.

In summary, the Pierre Robin Sequence of retrognathia, midline cleft palate and glossoptosis arises as a result of acute forward flexion of the head onto the chest in early pregnancy. It occurs once in around every 10,000 births. Provided that no other malformations are present, the prognosis is good.

### REFERENCES

1. Dunn, P.M. The influence of intrauterine environment in the causation of congenital postural deformities, with special reference to congenital dislocation of the hip. Thesis for MD Degree, University of Cambridge, 1969, Volume I, pp.160-161.
2. Dunn, P.M. Congenital postural deformities. *Brit. Med. Bull.*, 1976; 32, 71-76.
3. Davis, A.R. and Dunn, R. *Amer. J. Dis. Child.*, 1933, 45, 799.
4. Smith, D.W. Recognizable patterns in human deformation. Philadelphia. W.B. Saunders, 1981, pp.90-91 and 140.
5. Maas, C. and Poets, C.F. Initial treatment and early weight gain of children with Robin Sequence in Germany: a prospective epidemiological study. *Arch. Dis. Child. Fetal Neonatal Ed.*, 2014; 99, F491-F494.
6. Poswillo, D. The aetiology and surgery of cleft palate with micrognathia. *Ann. R. Coll. Surg. Eng.*, 1968; 43, 61.

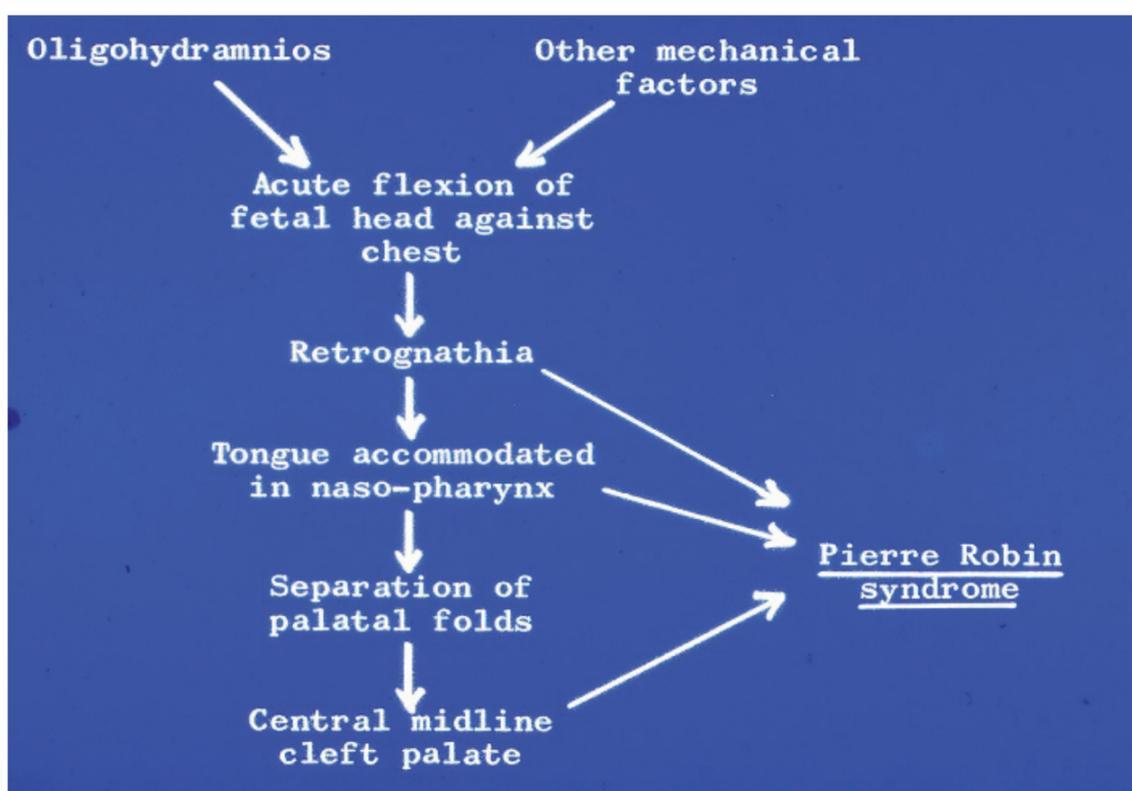


Figure 4  
Diagram of Pierre Robin Sequence.