Sindrom pierre robin: tata laksana multidisplin dan pengaruh terhadap luaran kasus

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Intisari

Latar belakang: Sindrom Pierre Robin atau terkadang disebut dengan *Pierre Robin Sequence* merupakan suatu kelainan kongenital yang ditandai dengan adanya trias *micrognatia, glossoptosis* serta *palatoskizis.* Penyebab pasti terjadinya sampai saat ini belum diketahui, tetapi beberapa kelainan genetik dihubungkan dengan kejadian sindrom pierre robin. Kejadian ini dapat berisfat *isolated* atau sindromik, pasien pasien dengan kelainan sindromik dapat terjadi kelainan pada organ lain. Tata laksana pada pasien membutuhkan pendekatan multidisiplin, karena adanya kompleksitas gejala serta memerlukan tindakan dari beberapa ahli.

Tujuan: Mengetahui efek tata laksana multidisiplin terhadap luaran anak dengan sindrom pierre robin

Metode: dilakukan pengamatan selama 18 bulan terhadap pasien berusia 2 tahun, yang didiagnosis dengan sindrom pierre robin, gizi buruk tipe marasmik, defek septum atrium, *profound sensorineural hearing loss*, dan global developmental delayed. Pasien dilakukan pengamatan dan mendapatkan intervensi multidisplin. Kemudian dilihat perbedaan pada awal dan akhir pengamatan.

Hasil: selama pengamatan kondisi pasien mengalami perbaikan, status nutrisi membaik dari gizi buruk menjadi gizi kurang, anak sudah bisa berjalan, terdapat periode pneumonia ringan dan tidak memerlukan perawatan inap, defek ASD menyempit spontan dan terjadi perbaikan kualitas hidup dilihat dari peningkatan nilai PedsQL.

Kata kunci: Sindrom pierre robin, tata laksana multidisiplin, kualitas hidup

Pierre robin syndrome: multi discipline approach and its effect on outcome

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Abstract

Background: Pierre Robin Syndrome (PRS) or sometimes referred to as the Pierre Robin Sequence is a congenital disorder characterized by a triad of micrognathia. glossoptosis and palatoschisis. The exact cause of PRS is unknown, but several genetic disorders have been associated with PRS, such as Stickler's syndrome or deletion 22q11 syndrome. In patients with PRS can be isolated or syndromic, PRS patients with syndromic disorders can experience other disorders, including heart defects, abnormalities in the hearing system, and growth and development disorders. The management of patients with PRS requires a multidisciplinary approach, because of the complexity of the symptoms and requires action from several experts.

Objective: To determine the effect of multidisciplinary management on the outcome of children with Pierre Robin syndrome.

Methods: 18-month observation was conducted on a patient aged 2 years, diagnosed with PRS, marasmus malnutrition, atrial septal defect, SNHL profound, and global developmental delay. Patients were observed and received multidisciplinary interventions from Plastic surgeon, ENT, Medical Rehabilitation, Social Pediatrics and Growth and Development, Nutrition and Metabolic diseases, and Pediatric cardiology. Then compare at the differences at the beginning and the end of the observation.

Results: during the observation that the patient's condition had improved clinically, the nutritional status improved from severe marasmus malnutrition to moderate malnutrition, the child was able to walk, there was a period of mild pneumonia but did not require hospitalization, ASD defects narrowed spontaneously and there was an improvement in the quality of life seen from the increase in the PedsQL value.

Key words: Pierre Robin syndrome, multidisciplinary management, quality of life