

Karakteristik klinis dan luaran terapi bisfosfonat pada pasien osteogenesis imperfekta di RS Cipto Mangunkusumo = Clinical features and outcome of bisphosphonate therapy in osteogenesis imperfecta patient at Cipto Mangunkusumo Hospital

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Abstrak

Osteogenesis imperfekta (OI) merupakan adalah penyakit genetik kelainan jaringan ikat berupa kerapuhan tulang dan fraktur berulang tanpa adanya trauma yang signifikan. Terdapat berbagai karakteristik klinis yang khas untuk mendiagnosis OI. Terapi bisfosfonat merupakan terapi utama pada OI yang bermanfaat untuk menurunkan insiden patah tulang agar tercapai kualitas hidup yang lebih baik. Penelitian ini bertujuan mengetahui karakteristik klinis dan luaran terapi bisfosfonat pada pasien anak dengan OI di RSCM. Penelitian ini dilakukan secara potong lintang terhadap 71 pasien OI berusia 0-18 tahun di RSCM pada 16-22 November 2020. Data diambil melalui kuesioner daring yang diisi oleh orangtua atau wali. Karakteristik klinis OI mencakup sklera biru (83%) dan patah tulang pada 69 (97%) pasien dengan lokasi paling banyak di tulang femur (66,2%). Hanya terdapat 18 subyek yang sudah melakukan pemeriksaan pendengaran dengan 4 (22%) diantaranya terdapat gangguan pendengaran. Klasifikasi klinis OI paling banyak adalah tipe berat (57%). Enam puluh lima subyek mengalami patahtulang di usia kurang dari 6 tahun termasuk intrauterin dan perinatal. Sebanyak 95,8% subyek mendapatkan terapi bisfosfonat dan hampir seluruhnya diberikan rutin setiap 6 bulan. Terdapat penurunan median kejadian patah tulang sebelum terapi bisfosfonat sebanyak 3,5 kejadian/tahun menjadi satu kejadian/tahun setelah terapi bisfosfonat. Terapi bisfosfonat dapat menurunkan angka kejadian patah tulang setiap tahun.

.....Osteogenesis imperfecta (OI) is a genetic disorder of connective tissue causing bone fragility and fractures in the absence of significant trauma. There are many typical clinical features of OI. Bisphosphonate therapy is the main therapy which significantly decreases fracture rate for better quality of life. This study was aimed to observe the clinical features and outcomes of bisphosphonate therapy in pediatric OI patients. A cross sectional study was conducted at Cipto Mangunkusumo Hospital (CMH) in the period of November 16th-22nd 2020. There were 71 patients aged 0-18 years old included for study analysis. Data were obtained from online questionnaire which was filled by their parents or guardians. The clinical features observed were blue sclera (83%) and fractures which occurred in 69 (97%) patients with the most common location was femur (66.2%). There were only 18 patients who underwent hearing examination and 4 of them (22%) had hearing problem. Most patients had severe OI classification (57%). Sixty-five patients had first fracture when their age <6 years old, including intrauterine and perinatal fractures. A total of 95.8% patients had received bisphosphonate therapy and almost all of patients had received treatment every 6 month. There was a decrease in the number of fractures from 3.5 events/year (before bisphosphonate therapy) to 1 event/year after bisphosphonate therapy. The outcome of bisphosphonate therapy was significant in terms of fracture incidence reduction. Bisphosphonate therapy was able to reduce fracture incidence per year.