

ABSTRAK

Latar Belakang: Sindrom Marfan adalah suatu penyakit genetik berupa kelainan jaringan ikat yang disebabkan oleh mutasi gen *FBN1*. Penyakit ini menyebabkan kondisi abnormal pada beberapa sistem organ, meliputi kardiovaskular, okular, muskuloskeletal, dan mungkin juga sistem organ lainnya. Penelitian ini dilakukan di RSUP Dr. Kariadi Semarang untuk memahami variasi klinis pada pasien sindrom Marfan.

Tujuan: Mengetahui frekuensi berdasarkan jenis kelamin, usia, serta variasi gejala klinis pasien dengan sindrom Marfan di RSUP Dr. Kariadi Semarang.

Metode: Data diperoleh dari rekam medik pasien RSUP Dr. Kariadi Semarang, dinilai berdasarkan kriteria inklusi dan eksklusi, dan dianalisis menggunakan Microsoft Excel dalam bentuk tabel.

Hasil: Penelitian deskriptif retrospektif dari 1 Januari 2018 - 31 Desember 2025 mengidentifikasi 8 pasien dengan sindrom Marfan. Pasien dengan usia terbanyak yaitu pada usia ≤ 20 tahun dengan 7 pasien dan didapatkan 7 pasien laki-laki dan 1 pasien perempuan. Berdasarkan sistem organ, pasien mengalami masalah muskuloskeletal (7 pasien), masalah okular (8 pasien), masalah kardiovaskular (4 pasien) dan masalah integumen (1 pasien). Pada masalah kardiovaskular didapatkan 1 pasien mengalami diseksi aorta, 1 pasien mengalami prolaps katup mitral, 2 pasien mengalami regurgitasi aorta, dan 3 pasien mengalami regurgitasi mitral. Pada masalah okular didapatkan 8 pasien mengalami miopia dan 5 pasien mengalami *ectopia lentis*. Pada masalah muskuloskeletal didapatkan 4 pasien dengan *dolichostenomelia*, 6 pasien dengan *arachnodactyly*, 6 pasien mengalami skoliosis thorakolumbal, dan 4 pasien mengalami deformitas dinding dada. Pada masalah integumen 1 pasien didapatkan mengalami striae kulit. Tidak ditemukan data pasien dengan masalah respirasi dan sistem saraf pusat.

Kesimpulan: Penelitian ini berhasil mengidentifikasi variasi klinis pasien sindrom Marfan di RSUP Dr. Kariadi selama 2018-2025, menemukan kelompok usia terbanyak ≤ 20 tahun, paling banyak berjenis kelamin laki-laki, dan pasien paling banyak mengalami masalah okular berdasarkan sistem organ. Pasien paling banyak mengalami masalah kardiovaskular yaitu regurgitasi mitral, masalah okular yaitu miopia, masalah muskuloskeletal yaitu *arachnodactyly* dan skoliosis thorakolumbal, dan masalah integumen yaitu striae kulit. Tidak didapatkan data pasien dengan masalah respirasi dan sistem saraf pusat.

Kata Kunci: Sindrom Marfan, karakteristik pasien, variasi klinis, RSUP Dr. Kariadi Semarang.

ABSTRACT

Background: Marfan syndrome (MFS) is a genetic disorder affecting connective tissue, caused by mutation in the *FBN1* gene. This condition leads to abnormalities in several organ systems, including the cardiovascular, ocular, and musculoskeletal systems, and potentially other organ systems. This study was conducted at Dr. Kariadi Hospital Semarang to understand the clinical variations in patients with MFS.

Aims: To determine the frequency based on age, gender and the variation of clinical symptoms in patients with MFS at Dr. Kariadi Hospital Semarang.

Methods: Data were obtained from the medical records of patients at Dr. Kariadi Hospital Semarang, assessed based on inclusion and exclusion criteria, and analyzed using Microsoft Excel in tabular form.

Results: A retrospective, descriptive study covering the period from January 1, 2018, to December 31, 2025, identified 8 patients with MFS. The largest age group was patients aged ≤ 20 years (7 patients), and the gender distribution consisted of 7 males and 1 female. Regarding organ systems, patients presented with musculoskeletal issues (7 patients), ocular issues (8 patients), cardiovascular issues (4 patients), and integument issues (1 patient). Cardiovascular findings included aortic dissection (1 patient), mitral valve prolapse (1 patient), aortic regurgitation (2 patients), and mitral regurgitation (3 patients). Ocular findings included myopia (8 patients) and ectopia lentis (5 patients). Musculoskeletal findings included dolichostenomelia (4 patients), arachnodactyly (6 patients), thoracolumbar scoliosis (6 patients), and chest wall deformity (4 patients). Integumentary findings included skin striae (1 patient). No data were found regarding respiratory or central nervous system issues.

Conclusion: This study successfully identified the clinical variations among MFS patients at Dr. Kariadi Hospital between 2018 and 2025; the findings revealed that the largest age group was ≤ 20 years old and the majority of patients were male. Regarding organ systems, ocular issues were the most prevalent. The most common conditions identified were mitral regurgitation (cardiovascular issues), myopia (ocular issues), and arachnodactyly and thoracolumbar scoliosis (musculoskeletal issues), and skin striae (integument issues). No data were obtained regarding patients with respiratory or central nervous system issues.

Keywords: *Marfan syndrome, patient characteristics, clinical variations, Dr. Kariadi Hospital Semarang.*