

## ABSTRAK

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### Latar belakang

Defek *radial ray* adalah salah satu penyakit genetik yang sebagian besar melibatkan anomali kongenital multipel. Terdapat 30 jenis anomali kongenital multipel yang memiliki tanda defek *radial ray*, antara lain Sindrom IVIC dan Anemia Fanconi. Kedua sindrom ini seringkali sulit dibedakan karena tandanya tumpang tindih. Membedakan Sindrom IVIC dengan Anemia Fanconi penting dilakukan karena memiliki perbedaan dalam hal perjalanan penyakit, prognosis kehidupan, prognosis disabilitas, dan pola pewarisan. Dengan mengamati pasien selama satu tahun atau lebih diharapkan dapat melihat tanda-tanda yang muncul secara berurutan sehingga bisa ditegakkan diagnosisnya.

### Kasus

Pasien laki-laki defek *radial ray* dan kelainan jantung, diamati setiap bulan berturut-turut selama satu tahun mulai usia 2 tahun 11 bulan. Sebelumnya, pasien juga diamati setiap kali periksa ke poliklinik anak sejak berusia 1 tahun 3 bulan. Terdapat riwayat dua kali keguguran. Saat awal pengamatan, pasien disuspek sebagai Sindrom IVIC. Dalam pengamatan didapatkan bahwa pasien mengalami mikrosefali dan gangguan pertumbuhan pascanatal. Inteligensi dan perkembangan motorik kasar untuk ekstremitas bawah baik. Pasien mengalami trombositopenia persisten yang semakin lama semakin berat. Trombositopenia diikuti oleh leukopenia. Atas tanda-tanda ini, diagnosis Anemia Fanconi ditegakkan, sedangkan diagnosis Sindrom IVIC gugur. Tidak didapatkan tanda-tanda infeksi berat, tetapi muncul tanda perdarahan spontan. Secara klinis tidak didapatkan kelainan hormonal yang berupa hipotiroidisme, hipokortisolisme, dan hipogonadisme. Kondisi pasien diterima dengan baik oleh orang tua, saudara, dan teman sebaya.

### Simpulan

Pasien dengan defek *radial ray* perlu dicurigai kemungkinan menderita Anemia Fanconi. Mikrosefali dengan inteligensi baik, gangguan pertumbuhan pascanatal, dan kelainan jantung bawaan termasuk tanda-tanda Anemia Fanconi. Kelainan



hematologis awal pada Anemia Fanconi muncul secara berurutan, yaitu trombositopenia, diikuti oleh leukopenia.

**Kata kunci**

Anemia Fanconi; Sindrom IVIC; radial ray; trombositopenia; anemia aplastika

## ABSTRACT

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### Backgorund

Radial ray defects are genetic diseases that many of them are part of multiple congenital anomalies. There are 30 kinds of multiple congenital anomalies that signs consist of radial ray defect, e.g. IVIC Syndrome and Fanconi Anemia. Both syndromes are often difficult to be differentiated because of their overlapping signs. Differentiating IVIC Syndrome from Fanconi Anemia is important because both of them have different disease courses, prognoses, and mode of heredity. By observation for one year or more, it is expected to see signs that appear simultaneously, so the diagnosis can be established.

### Case

A male patient with radial ray defect and congenital heart disease, was observed every months sequentially for one year since 2 years 11 months old of age. Previously the patient was observed everytime the patient was brought by his parents to pediatric clinic for routine examination since 1 year 3 months old of age. There were history of miscarriages twice. On the early observation, patient was suspected as IVIC Syndrome. On observation, it was found that the patient had microcephaly, and suffered from postnatal growth retardation. Intelligence and gross motoric development of lower extremities was normal. There was persistent thrombocytopenia followed by leukopenia. Because of these signs, diagnosis of Fanconi Anemia was established. Diagnosis of IVIC Syndrome was dropped. No signs of severe infections. There were signs of spontaneous hemorrhage. Clinically no signs of hormonal abnormalities, i.e. no hypothyroidism, no hypocortisolism, and no hypogonadism. Patient's physical disabilities could be accepted well by parents, families, and friends.



## **Conslusions**

Patients suffering from radial ray defect should be suspected as Fanconi Anemia. Microcephaly with normal intelligence, postnatal growth retardation, and congenital heart defect were also signs of Fanconi Anemia. Its hematologic abnormalities appeared sequentially, i.e. thrombocytopenia, followed by leukopenia.

## **Keywords**

Fanconi Anemia; IVIC Sybndroma; radial ray; thrombocytopenia; aplastic anemia.