Family profile of Down syndrome patients in West Java Province, Indonesia

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Abstract

Down syndrome is the most common chromosomal abnormality characterised by complete chromosome 21 trisomy (classical), or partial chromosome 21 trisomy (mosaicism), or chromosome rearrangement involving chromosome 21 (translocation). The study was conducted to describe the family and social profile of Down syndrome patients who were referred to a research centre in Bandung, the capital city of West Java Province, Indonesia. This descriptive study provides picture of residential location and surrounding area of their dwellings around West Java Province region, which previous studies showed it as risk factors. This study involved people with Down syndrome in various ages whose chromosomes were examined by conventional karyotyping and Fluorescent In-Situ Hybridization (FISH) method. Data were collected from 205 patients with Down syndrome, during the period of September 2015 to March 2018, who were referred to Cell Culture and Cytogenetics Laboratory, Faculty of Medicine Universitas Padjadjaran, Indonesia. Results showed that the most common type of Down syndrome was classical 195 patients (95%), followed by mosaic 6 patients (3%) and translocation 4 patients (2%). Most of them live in urban area 84 patients (41%) while the rest live in the suburban area 76 patients (37%), and only a few came from rural area 15 patients (7%) and the rest were unknown area or uncompleted data. They live mostly in dense area (51%), in mid-density area (31%) and the rest live in sparse populated area (18%). Their houses are mostly located in residential estate area 107 patients (52%), nearby industrial area 38 patients (18%), and around agricultural area 30 patients (15%). For maternal age, there were 84 mothers (40%) with 35 years old and more, and the rest under 35 years old. Based on the study, it could be concluded that classical Down syndrome is the most common type, where the majority live in dense residential area. This study offered several new information regarding frequency, average age, area of Down syndrome patients’ residence and the condition of neighbourhood in West Java where they live.

Keywords: Chromosome; descriptive; Down syndrome; prevalence

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