Prevalence of Down Syndrome: A Single-Center Study in Bandung, Indonesia

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Abstract : Down syndrome (DS) is a chromosomal abnormality characterised by complete 21 chromosome trisomy (classical or non-disjunction), or partial 21 chromosome trisomy (mosaicism), or chromosome rearrangement involving chromosome 21 (translocation). This study was carried out to describe the frequency of DS patients in a research institution in the city of Bandung, Indonesia. This descriptive study also provides a picture of the residential location and surrounding area of their dwellings. This study involved people with DS in various age whose chromosome were evaluated by conventional karyotyping method and FISH. Data were collected from 60 patients with DS from a total 150 patients during the period of September 2015 to August 2016 who were referred to Cell Culture and Cytogenetics Laboratory, Faculty of Medicine Universitas Padjadjaran, Indonesia. Results showed that the most common type of DS was non-disjunction (93%), followed by mosaicism (5%), no patient with translocation DS (0%), and a very rare type of tetrasomy 21 (2%). There were 39 males (65%) and 21 females (35%) of DS patient. Most of them live in suburban area beyond Bandung city (55%) while the rest live inside urban area of Bandung city (45%). They live mostly in dense area of greater Bandung area (65%) and only a few live in mid-density area (25%) and the least live in sparse populated area (10%). Their houses are mostly located in residential estate area (55%), nearby industrial area (37%), and around agricultural area (8%). Based on the study, it could be concluded that non-disjunction DS is the most common type. DS patients referred to the laboratory mostly came from dense residential zone in suburban area outside Bandung city. The low number of DS patients referred to the laboratory for chromosome analysis was the highlight to improve health service for people with genetic disorder. This study offered several information regarding area of DS patients' residence and the condition of neighbourhood in Bandung city where they live as well.

Keywords : chromosome, descriptive, Down syndrome, prevalence

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