I. Introduction

Genetic disorder is a condition caused by the alteration of hereditary material, the DNA, away from the normal sequence. As a blueprint of life, the alteration of the DNA in every single structure start from nucleotide, the gene, and the chromosome might cause some serious medical condition such as morphological change of the body, mental retardation, and dysfunction of organs. Genetic disorder classified into three main categories and global prevalence for each categories was single gene disorder (3.6 per 1000), chromosomal abnormalities (1.8 per 1000), and multifactorial inheritance (46.4 per 1000).

Locally, in Indonesia, the genetic disorders prevalence data still remain unknown except for some specific genetic disorder such as Down Syndrome (1.2 per 1000). Further study about genetic disorders prevalence data is needed to help the authorities to build right management for other genetic disorders. This study aimed to determine genetic disorders prevalence in Cytogenetics Laboratory, Faculty of Medicine, Universitas Padjadjaran, during 2015-2016.

II. Subject and Method

165 patients with suspicion of having genetic disorder, examined using Karyotype and FISH. Classified the result into 2 main categories: Chromosomal Abnormalities and Other Genetic Disorder. Descriptive and Frequency Distribution Analysis.

III. Results and Discussion

Results showed that the genetic disorders prevalence in Cytogenetics Laboratory, Faculty of Medicine, Universitas Padjadjaran, during 2015-2016 were 53.33% for chromosomal abnormalities and 46.47% for other genetic disorder (Figure 1). The abnormalities of chromosome could be identified as structural abnormalities (translocation, deletion, insertion, etc.) or numerical abnormalities (monosomy, trisomy, tetrasomy, and mosaic) and other genetic disorder could be classified into four sub-categories as normal XY, normal XX, disorder of sexual development (DSD) XY, DSD XX, and other unidentified genetic abnormalities.

![Figure 2](image1)

The prevalence of each subcategory were monosomy (4.55%), trisomy (88.64%), tetrasomy (1.14%), translocation (1.14%), and mosaicism (4.55%) (Figure 2). Trisomy has the highest prevalence of chromosomal abnormalities and the most common incidence is trisomy 21, which caused Down syndrome. The other syndrome identified in this study, which caused by trisomy were Patau (+13), Edward syndrome (+18), and Kleinfelter syndrome (XXY). In monosomy sub-category, patient was identified as Turner syndrome (XO), while in tetrasomy case there were 2 extra chromosome 21 which also caused Down syndrome. Based on the data in this study, there were two incidences of mosaicism, XO/XX case and XY/XY +21.

IV. Conclusion

The prevalence of genetic disorders in Cytogenetics Laboratory, Faculty of Medicine, Universitas Padjadjaran during 2015-2016 was 53.33% for chromosomal abnormalities, while the prevalence of other genetic disorder is 46.67%.

V. References

Genetic Disorders Prevalence in Cytogenetics Laboratory, Faculty of Medicine, Universitas Padjadjaran in 2015-2016

Rosyada, Nur Amalia; Laksono, Bremmy; Parikrama, Riksa; Suciati, Lita Putri; Effendi, Dadang Sjarief Hidajat
The Center of Genetic Study, Faculty of Medicine, Universitas Padjadjaran, Indonesia; rosyadanuramalia@gmail.com

Genetic disorder is a condition caused by the alteration of hereditary material, the DNA, away from the normal sequence. The alteration of DNA in every single structure start from the nucleotide, the gene, and the chromosome might cause some serious medical condition such as morphological changes of the body, mental retardation, and dysfunction of organs. The statistical information about genetic disorder such as prevalence data is needed to help the authorities find the right management for the diseases. The aim of this study is to determine genetic disorder prevalence in Cytogenetics Laboratory, Faculty of Medicine, Universitas Padjadjaran during 2015 to 2016. Data were collected from chromosomal analysis of 165 Cytogenetic Laboratory patients during period of 2015 to 2016. Descriptive and frequency distribution analysis were performed to obtain the genetic disorder prevalence. Result showed that 53.3% patients have chromosome abnormalities while 46.7% patients have other genetic disorders. In chromosome abnormalities category, the prevalence was monosomy (4.55%), trisomy (88.64%), tetrasomy (1.14%), chromosome translocation (1.14%), and mosaicism (4.55%). Meanwhile, the prevalence of other genetic disorder was male patients with normal chromosome (55.84%), female patients with normal chromosome (35.06%), male patients with disorder of sexual development (5.19%), female patients with disorder of sexual development (0%), and other abnormalities (3.9%). Based on the results, it can be concluded that the prevalence of chromosome abnormalities is higher than other genetic disorders. Therefore, the awareness about genetic disorder, not only chromosome abnormalities but also other type disorders, need to be increased in community and particularly for clinicians.

Keyword: genetic disorders, chromosomal abnormalities, prevalence