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

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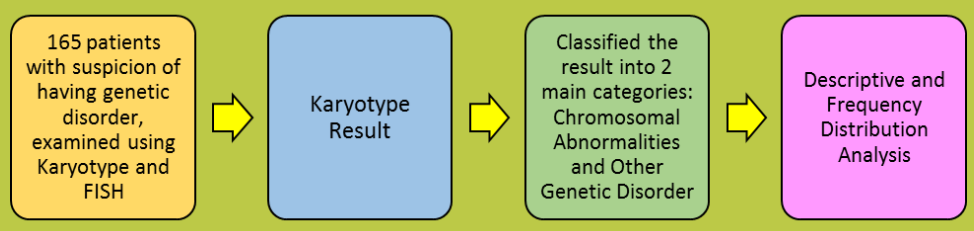
I. Introduction

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.[1] 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).[2] Locally, in Indonesia, the genetic disorders prevalence data still remain unknown except for some spesific genetic disorder such as Down Syndrome (1.2 per 1000)[3]. 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 Padjajaran, during 2015-2016.

II. Subject and Method



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.

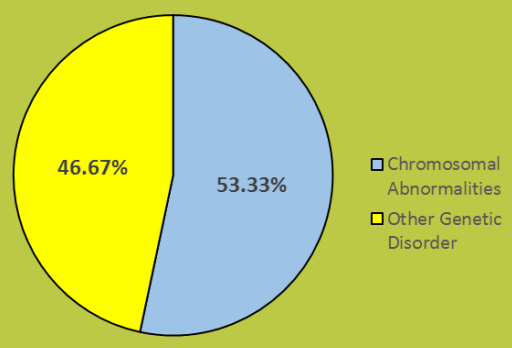


Figure 1. Genetic disorder prevalence in Cytogenetic Laboratory, Faculty of Medicine, Universitas Padajajaran during 2015-2016. Chromosomal abnormalities prevalence (showed as blue) was 53.33% while other genetic disorder (showed as yellow) was 46.67%.

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.[4] Based on the data in this study, there were two incidences of mosaicism, XO/XX case and XY/XY +21 .

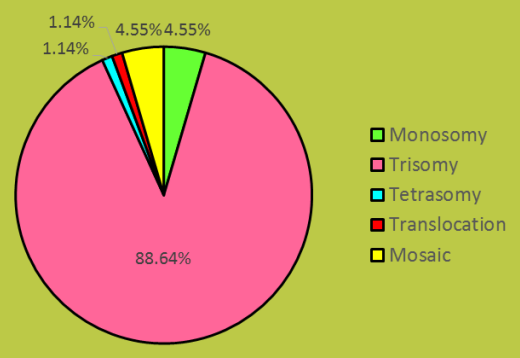


Figure 2. Pie chart of chromosomal abnormalities prevalence in Cytogenetic Laboratory, Faculty of Medicine, Padajajaran University during 2015-2016. The prevalence for each categories was Monosomy (4.55%), Trisomy (88.64%), Tetrasomy (1.14%), Translocation (1.14%), and Mosaic (4.55%).

Although the genetic disorders were classified into three main categories as chromosomal abnormalities, single gene disorder, and multifactorial disorder, the last two category could not be differentiated because it needs further detailed examination such as DNA analysis. Therefore, they were categorized as other genetic disorder. Other genetic disorders were then divided into sub-categories based on the case of incidence. The prevalence for each sub-categories were normal XY (55.84%), normal XX (35.06%), DSD XY (5.19%), DSD XX (0%), and other abnormalities 3.9%) (Figure 3). The most common incidence was normal XY. The karyotype result showed no abnormalities in the chromosome, although the physical appearence of the patient showed a spesific DSD characteristic such as micropenis and hypospadias.[5]

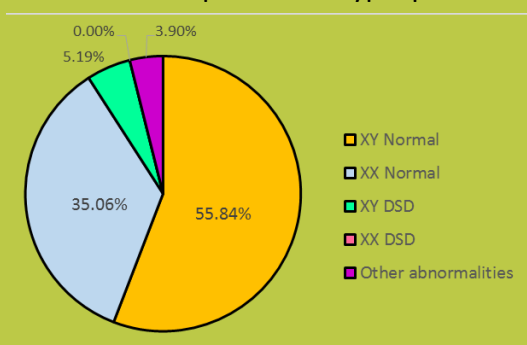


Figure 3. Other genetic disorder prevalence in Cytogenetic Laboratory, Faculty of Medicine, Universitas Padjadjaran during 2015-2016. The prevalence for each categories were Normal XY (55.84%), Normal XX (35.06%), DSD XY (5.19%), DSD XX (0%), and other abnormalities (3.90%).

IV. Conclusion

The prevalence of genetic disorders in Cytogenetic 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

1. Mochtar. 1997. Chromosomal aberration in children with suspected genetic disorder. *Estern Mediteranian Health Journal* 3(1): 114-122.

2. Rimon DL, Connor JM, Pyeritz. 1997. Principles and practices of medical genetics. New York.

3. KEMENKES RI. 2013. Riset Kesehatan Dasar 2013. Jakarta: Badan Penelitian dan Pengembangan Kesehatan Kemenkes RI.

4. Wynbrandt, J. 2008. The encyclopedia of genetic disorder and birth defects, third edition. New york: Fact on File, Infobase Publishing.

5 Ocal, G. 2011. Current concepts in Disorders of Sexual Development. J Clin Res Pediatr Endocrinol 3(3): 105-114

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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