

P5

PHENOTYPE AND CYTOGENETIC PROFILE OF DOWN SYNDROME CASES IN SPECIAL SCHOOLS IN SEMARANG

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Down syndrome (DS) is the most common genetic cause of mental retardation. This disease is caused by chromosomal aberration, characterized by well-defined and distinctive phenotypic features with very complicated dimorphology. The diagnosis of DS should be established by cytogenetic examination, other than this test is expensive. Alternatively physical assessment and some categories in making early clinical diagnosis of DS should be considered. This study was designed to identify physical characteristics and cytogenetic profile in Down syndrome in special schools (for children with special needs) in Semarang and made early clinical diagnosis of DS.

This study used a descriptive observational method. Samples from four special schools in Semarang were selected using Down syndrome characteristics checklist during period of July 2007 until July 2008. Forty five heparinised bloods were collected for cytogenetic analyses, five samples have been examined previously in the course of diagnostic. There were 45 students with positive trisomy 21 and 5 students with normal karyotype, and the clinical examination accuracy was 90%. Down Syndrome characteristics were upslanting palpebra fissure (76%), flat facial profile 94%, ear abnormality 72%, hypotonia 64%, simian crease 56%, saddle sign 80%, hypertelorism 46%, short stubby fingers 56%, protruding tongue 36%, clinodactily 56%, epicanthic folds 90%, and excessive skin fold on neck 12%. Forty five samples were trisomy 21 and five samples did not have structural and numeral abnormality. There were twelve physical characteristics that can be used to make early clinical diagnosis of Down syndrome, with accuracy 90%.

Keywords: *Down syndrome clinical sign, Cytogenetics*

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