

Faktor genetik dan non genetik pada manusia pigmi Rampasasa Flores = Genetic and non genetic study in human pygmies Rampasasa Flores

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Abstrak

[ABSTRAK

Latar belakang: Tinggi badan/ perawakan tubuh merupakan parameter penting tingkat kesejahteraan suatu populasi. Perawakan tubuh dipengaruhi oleh faktor genetik, endokrin dan lingkungan. Faktor lingkungan yang saat ini paling sering ditemukan adalah faktor nutrisi. Populasi pigmi adalah suatu populasi terisolasi yang seluruh anggotanya pendek dan ditemukan di berbagai belahan dunia termasuk Indonesia, yaitu di Flores, Nusa Tenggara Timur yang disebut komunitas pigmi Rampasasa. Sampai saat ini belum ada penelitian yang dapat menemukan penyebab perawakan pendek komunitas pigmi tersebut.

Tujuan: Mengetahui profil antropometri manusia pigmi Rampasasa dan mencari berbagai faktor (genetik, endokrin, dan nutrisi) yang berperan dalam perawakan pendek komunitas pigmi tersebut sehingga diharapkan dapat berkontribusi dalam tatalaksana perawakan pendek pada umumnya.

Metode: Penelitian merupakan studi deskriptif analitik yang dilakukan pada periode Desember 2011-April 2014. Penelitian ini menggunakan desain potong lintang untuk mengetahui profil genetik dan non genetik (endokrin dan nutrisi) yang berperan dalam perawakan pendek manusia pigmi Rampasasa. Dilakukan pengukuran antropometri pada subjek dan pengambilan sampel darah. Analisis statistik dilakukan dengan uji ANOVA yang dilanjutkan dengan post hoc analysis. Analisis genetik dilakukan dengan mengirimkan isolasi DNA ke Laboratory for Diagnostic Genome Analysis (LDGA), Leiden, Belanda.

Hasil: Didapatkan data dari 58 subjek yang dikelompokkan menjadi pigmi murni (n=8), pigmi campuran (n=40), dan non pigmi (n=10). Seluruh subjek memiliki proporsi tubuh yang normal. Tidak terdapat perbedaan bermakna untuk status nutrisi antara ketiga kelompok, yang dinyatakan dengan kadar kalsium (p=0,19), vitamin D (p=0,96), dan hemoglobin (p=0,147). Namun didapatkan perbedaan bermakna untuk kadar hormon IGF-1 antara ketiga kelompok (p=0,037), yang setelah dilakukan analisis posthoc menunjukkan perbedaan hanya pada kelompok non pigmi vs. pigmi murni (p=0,012). Kadar hormon IGFBP-3 tidak menunjukkan perbedaan bermakna antara ketiga kelompok (p=0,772). Analisis DNA menggunakan SNP array mengidentifikasi 10 regio homozigot pada sampel pigmi yang tidak didapatkan pada kontrol.

Simpulan: Perawakan pendek manusia pigmi Rampasasa memiliki proporsi tubuh yang normal. Faktor nutrisi tidak berhubungan dengan perawakan pendek komunitas pigmi Rampasasa. Faktor hormonal tidak dapat menjelaskan perawakan pendek populasi tersebut. Temuan regio homozigot mengindikasikan pengaruh faktor genetik meskipun kandidat gen belum dapat diidentifikasi.;

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ABSTRACT

Background: Height / stature of the body is an important parameter of one?s population wellbeing. Height is influenced by genetic, endocrine and environmental factors. Nutritional factor is one of the most common environmental factors found. Pygmy population is an isolated population whose all members have short

stature. They are found in various parts of the world including Indonesia, namely Rampasasa pygmies community in Flores, East Nusa Tenggara. Up until this date, there are no studies about the etiology of Rampasasa pygmies short stature.

Objective: To learn about anthropometric profiles in Rampasasa Pygmies and factors involved in the short stature of that pygmy community, as a contribution to the management of short stature in general, as well as to provide scientific asset about Rampasasa pygmies.

Keywords: calcium, genetic factors, height, IGFBP-3, IGF-I, nutrition, Rampasasa pygmies, short stature, vitamin D.

Methods: This research is a descriptive analytic study conducted from December 2011 to April 2014. This study used a cross-sectional design to determine the genetic and non-genetic profile (endocrine and nutrition) that play role in Rampasasa pygmies short stature. Anthropometric measurements and blood sampling were performed. Statistical analysis was performed by using ANOVA followed by post hoc analysis. Genetic analysis is done by sending DNA isolation to Laboratory for Diagnostic Genome Analysis (LDGA), Leiden, The Netherlands.

Results: Data obtained from 58 subjects were grouped into pure pygmies (n = 8), mixed pygmy (n = 40), and non- pygmies (n = 10). All subjects had normal body proportions. There were no significant difference in nutritional status between three groups, which is expressed by calcium level (p = 0.19) , vitamin D (p = 0.96), and hemoglobin (p = 0.147). Significant difference of IGF-1 hormone were found between the three groups (p = 0.037), which after posthoc analysis showed differences only between non-pygmies vs. pure pygmies (p = 0.012). IGFBP-3 hormone level showed no significant difference among the three groups (p = 0.772). We obtained evidence of homozygous regions in DNA analysis using SNP arrays method, which are not found in control group.

Conclusion: Rampasasa pygmies have short stature with normal body proportion.

Nutritional factors are not associated with short stature of Rampasasa pygmy communities. Hormonal factors can not explain the cause of the population short stature.

The discovery of homozygous regions indicates the role of genetic cause even though

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