Studi epidemiologi variasi gen NDP (norrie disease pseudoglioma) dan faktor layanan neonatal pada penderita retinopati prematuritas = Epidemiology study of NDP (norrie disease pseudoglioma) gene variation and neonatal care on infants with retinopathy of prematurity

Johanes Edy Siswanto, author

Abstrak

Latar Belakang: Retinopati Prematuritas atau ROP merupakan gangguan vaskular retina bayi prematur yang dapat menyebabkan pelepasan retina dan terjadinya kebutaan. Variasi gen Norrie disease Pseudoglioma (NDP) serta paparan oksigen diduga terlibat dengan kejadian dan perkembangan ROP.

Tujuan: Mengetahui peran variasi NDP serta faktor layanan neonatal khususnya paparan oksigen dalam memprediksi kejadian ROP pada bayi prematur Indonesia. Metodologi: Studi dilaksanakan tahun 2009-2014 di beberapa pusat pelayanan perinatologi dan mata sekitar Jakarta. Sebanyak 6 situs mutasi pada ekson 3 dideteksi yaitu C597A, L108P, R121W, A105T, Val60Glu, dan C110G. Perubahan susunan basa gen NDP dianalisis dengan mengampilifikasi gen NDP bagian ekson 3 menggunakan metode Polymerase Chain Reaction (PCR-RFLP dan PCR-SSP). Hasil diverifikasi dengan sekuensing DNA. Model multivariat hasil analisis regresi logistik dan regresi Cox digunakan sebagai model skoring untuk memprediksi kejadian dan keparahan ROP.

Hasil: Tidak ditemukan polimorfisme dan mutasi pada situs NDP exon 3. Hasil analisis multivariat didapatkan BBL, PJT(NCB-KMK), transfusi tukar, lama suplementasi O2, SpO2 terendah, dan sosial ekonomi sebagai variabel yang berhubungan dengan kejadian ROP. Sedangkan dalam hubungannya dengan keparahan ROP, didapatkan usia gestasi, lama suplementasi O2 > 7 hari, SpO2 terendah, rujukan RS, dan sosial ekonomi.

Kesimpulan: Tidak didapatkan polimorfisme dan mutasi gen NDP exon 3 pada kasus ROP bayi prematur Indonesia. Lama suplementasi O2 dan nilai SpO2 terendah mempunyai peran dalam meningkatkan risiko kejadian dan berkembangnya ROP menjadi lebih berat.
<hr>
Background: Retinopathy Prematurity or ROP is retinal vascularization disorder on premature infants that causing retina detachment and eventually blindness. NDP gene mutation and oxygen exposure might have role in incidence of ROP.

Objective: This study was conducted to determine the role of NDP gene polymorphism and mutation, and oxygen exposure for predicting the incidence of ROP in Indonesia.

Methodology: Data were collected from few Perinatology and Opthalmology centres around Jakarta during 2009-2014. DNA samples isolated from blood and buccal cell. This study tried to detect 6 mutations site on exon 3 of NDP gen which are C597A, L108P, R121W, A105T, Val60Glu, and C110G. Alterations of NDP

gene were analized with amplification of NDP gene in exon 3 region using Polymerase Chain Reaction (PCR-RFLP dan PCR-SSP) methods. The result verified with DNA sequencing. Scoring model were made by using logistic regression to predict the incidence and development of ROP.

Result: No NDP gene polymorphism and mutations at exon 3 region was detected. The result have been analized with PCR-RFLP and verified with DNA sequencing. Multivariate analysis using logistic regression for incidence of ROP retain birth weight, IUGR, gender, respiratory distress, exchange transfussion, length of O2 supplementation, SpO2 minimum, and socioeconomic variables. As for ROP severity, multivariate analysis retain gestational age, gender, access to hospital (inborn/outborn), apnea, length of O2 supplementation, SpO2 minimum, and socioeconomic variables.

Conclusion: The relationship between polymorphisms and mutations of NDP gene and ROP cases that happened in Indonesian premature infants population did not showed in this study. Length of O2 supplementation and minimum value of SpO2 85 - 90% significantly increase the risk for ROP incidence and development of severe ROP.