

## ABSTRAK

**Latar belakang:** Prevalensi ASD meningkat dalam tiga dekade terakhir. Komponen etiologis genetik pada ASD cukup kuat dan kompleks. Diperkirakan terdapat 888 gen yang berpotensi terlibat pada kejadian ASD. Gen OXTR dan AVPR1B adalah dua gen yang dikaitkan dengan risiko ASD. Meskipun faktor genetik jelas berkontribusi terhadap risiko ASD, namun faktor lingkungan, *prenatal* dan *post-natal* juga terlibat.

**Tujuan:** Mengidentifikasi hubungan antara polimorfisme gen OXTR dan AVPR1B dengan ASD dan gangguan perilaku sosial, serta besar risiko dari faktor-faktor risiko yang berhubungan dengan kejadian ASD dan skor perilaku sosial.

**Metode:** Desain penelitian analitik molekuler dengan desain penelitian *case control*. Wilayah penelitian di Eks Karesidenan Banyumas. Besar sampel kelompok kasus 47 anak dan kontrol 56 anak. Instrumen meliputi data demografi, pengukur skor respons perilaku sosial, set alat-bahan untuk *sampling* darah. Analisis data menggunakan *chi square*, *independent t test*, *general linear model*, regresi logistik, regresi linear, *SNP-stat*.

**Hasil:** Terdapat perbedaan signifikan perilaku sosial anak pada variasi genotip G/A dan G/G gen OXTR rs2254298 ( $p = 0,04$ ). Terdapat perbedaan sangat signifikan antara skor perilaku sosial kelompok ASD dan kontrol ( $p < 0,001$ ). Faktor yang berhubungan signifikan dengan ASD yaitu jenis kelamin ( $p = 0,009$ ; OR = 3,05); usia gestasi ( $p = 0,043$ ; OR = 3,12); riwayat penyakit anak ( $p < 0,001$ ; OR = 5,91); gangguan selama kehamilan ( $p = 0,03$ ; OR = 2,4). Faktor-faktor yang mempengaruhi gangguan perilaku sosial anak meliputi riwayat sakit anak ( $p = 0,09$ ), paparan asap rokok pada saat kehamilan ( $p = 0,19$ ).

**Kesimpulan:** Terdapat perbedaan signifikan perilaku sosial variasi genotip G/A dengan G/G pada gen OXTR rs2254298. Terdapat perbedaan signifikan perilaku sosial kelompok ASD dengan kontrol. Anak laki-laki tiga kali lebih berisiko ASD dibanding perempuan, anak lahir prematur tiga kali lebih berisiko ASD, anak yang saat balita mengalami sakit enam kali lebih berisiko ASD, ibu yang masa kehamilannya mengalami gangguan kesehatan dua kali lebih berisiko anaknya ASD. Perilaku sosial dipengaruhi oleh riwayat sakit anak, dan paparan asap rokok pada ibu saat hamil.

**Kata kunci:** ASD, OXTR, AVPR1B, perilaku sosial

## ABSTRACT

**Background:** The prevalence of ASD has increased in the last three decades. The genetic aetiological component in ASD is quite strong and complex. It is estimated that there are 888 genes that are potentially involved in ASD events. The OXTR and AVPR1B genes are two genes associated with ASD risk. Although genetic factors clearly contribute to the risk of ASD, environmental, prenatal and post-natal factors are also involved.

**Objective:** To identify the relationship between OXTR and AVPR1B gene polymorphisms with ASD and impaired social behavior, as well as the risk of factors related to ASD events and social behavior scores.

**Methods:** Design of molecular analytic research with a case control and cross sectional research design. Research area in the former Residency of Banyumas. The sample size of the group was 47 children and controls 56 children. Instruments include demographic data, gauges of social behavior response scores, sets of tools for blood sampling. Data analysis using Chi square, independent t test, general linear model, logistic regression, linear regression, SNP-stat.

**Results:** There were significant differences in children's social behavior on G/A and G/G genotypes OXTR rs2254298 ( $p=0.04$ ). There was a very significant difference between the scores of social behavior in the ASD group and controls ( $p<0.001$ ). Factors that were significantly associated with ASD were gender ( $p=0.009$ , OR=3.05); gestational age ( $p=0.043$ , OR=3.12); history of childhood illness ( $p<0.001$ , OR=5.91); disorders during pregnancy ( $p=0.03$ , OR=2.4). Factors that influence the disruption of children's social behavior include a history of childhood illness ( $p=0.09$ ), exposure to cigarette smoke during pregnancy ( $p=0.19$ ).

**Conclusion:** There is a significant difference in the social behavior of G/A genotype variation with G/G on the OXTR gene rs2254298. There were significant differences in the social behavior of the ASD group with controls. Boys were three times more at risk of ASD than women, children born prematurely were three times more at risk of ASD, children who are ill when childhood 0-5 years old were six times more at risk of ASD, mothers who have a pregnancy with health problems were two times more likely to have ASD. Social behavior is influenced by a child's history of illness, and exposure to cigarette smoke in the mother during pregnancy.

**Keywords:** ASD, OXTR, AVPR1B, social behavior