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授与した学位	博　　士
専攻分野の名称	医　　学
学位授与番号	博甲第 3982 号
学位授与の日付	平成２１年９月３０日
学位授与の要件	医歯薬学総合研究科生体制御科学専攻 （学位規則第４条第１項該当）
学位論文題目	Chromosomes 4q28.3 and 7q31.2 as New Susceptibility Loci for Comitant Strabismus （共同性斜視関連染色体座位 4q28.3および7q31.2）
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学位論文内容の要旨

Strabismus is the misalignment of the eyes and can be classified as comitant when a deviation is constant in all directions of gaze, or incomitant when a deviation varies in different directions of gaze. The prevalence of comitant strabismus in Japan is approximately 1 % to 2% of the population and approximately 2% to 4% among Caucasian populations. Family, twin and population studies suggest a strong hereditary background for comitant strabismus. To date, only two genome-wide linkage studies of comitant strabismus have been published. The current study was designed to localize chromosomal susceptibility loci for comitant strabismus among Japanese families by genome-wide linkage analyses. Fifty-five Japanese families, with at least two members with comitant strabismus, were subject to full ophthalmic examination, ocular history, and review of medical records. DNA was obtained and genotyping was performed with PCR amplification of 400 micro satellite markers. Parametric and nonparametric linkage (NPL) analyses scores were calculated. Linkage analysis was performed for the whole set of families, and then a second analysis was performed for two subgroups with the phenotypes, esotropia and exotropia. A multipoint parametric heterogeneity logarithm of the odds (HLOD) score of 3.62 was obtained at marker D4S1575 under a dominant model, with a NPL score of 2.68 (*P*=0.001). Testing under different penetrances and disease allele frequencies revealed two other susceptibility loci at 7q31.2 under a recessive model (HLOD scores = 3.93 and 4.40 at 125.2 cM and 107.28 cM, respectively). Analysis of the subgroups revealed new susceptibility loci for esotropia; one locus at 8q24.21 is worthy of further investigation. This study suggests multiple susceptibility loci for comitant strabismus. The loci at chromosomes 4q28.3 and 7q31.2 show a significant evidence of linkage.

論文審査結果の要旨

本研究は,共同性斜視の病因として、従来報告されていた 7p22.1 ではなく、4q28.3 と 7q31.2 の染色体座位が関連することを岡山大学斜視外来の患者家族の連鎖解析によって解明した研究で、将来の共同性斜視の遺伝子診断や手術適応につながる重要な知見を示し，価値ある業績であると認められる。

よって，本研究者は博士（医学）の学位を得る資格があると認める。