

ACKNOWLEDGMENTS

This study was supported by the Health and Labour Sciences Research Grants of Research on Intractable Diseases from the Ministry of Health, Labour and Welfare, Tokyo, Japan.

REFERENCES

- Aramaki M, Ueda T, Kosaki R, Makita Y, Okamoto N, Yoshihashi H, Oki H, Nanao K, Moriyama N, Oku S, Hasegawa T, Takahashi T, Fukushima Y, Kawame H, Kosaki K. 2006a. Phenotypic spectrum of CHARGE syndrome with CDH7 mutations. *J Pediatr* 148:410–414.
- Aramaki M, Ueda T, Torii C, Samejima H, Kosaki R, Takahashi T, Kosaki K. 2006b. Screening for CHARGE syndrome mutations in the CDH7 gene using denaturing high-performance liquid chromatography. *Genet Test* 10:244–251.
- Blake KD, Davenport SL, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JM Jr. 1998. CHARGE association: An update and review for the primary pediatrician. *Clin Pediatr (Phila)* 37:159–173.
- Cohen J. 1960. A coefficient of agreement for nominal scales. *Educ Psychol Meas* 20:37–46.
- Holak HM, Kohlhase J, Holak SA, Holak NH. 2008. New recognized ophthalmic morphologic anomalies in CHARGE syndrome caused by the R2319C mutation in the CHD7 gene. *Ophthalmic Genet* 29:79–84.
- Hornby SJ, Adolph S, Gilbert CE, Dandona L, Foster A. 2000. Visual acuity in children with coloboma. Clinical features and a new phenotypic classification system. *Ophthalmology* 107:511–520.
- Jongmans MC, Admiraal RJ, van der Donk KP, Vissers LE, Baas AF, Kapusta L, van Hagen JM, Donnai D, de Ravel TJ, Veltman JA, van Kessel AG, De Vries BB, Brunnaer HG, Hoefsloot LH, van Ravenswaaij CM. 2006. CHARGE syndrome: The phenotypic spectrum of mutations in the CHD7 gene. *J Med Genet* 43:306–314.
- Lalani SR, Saliullah AM, Fernbach SD, Harutyunyan KG, Thaller C, Peterson LE, McPherson JD, Gibbs RA, White LD, Hefner M, Davenport SLH, Graham JM Jr, Bacino CA, Glass NL, Towbin JA, Craigen WJ, Neish SR, Lin AE, Belmont JW. 2006. Spectrum of CHD7 mutations in 110 individuals with CHARGE syndrome and genotype–phenotype correlation. *Am J Hum Genet* 78:303–314.
- Landis JR, Koch GG. 1977. The measurement of observer agreement for categorical data. *Biometrics* 33:159–174.
- Pagon RA, Graham JM Jr, Zonana J, Yong SL. 1981. Coloboma, congenital heart disease, and choanal atresia with multiple anomalies: CHARGE association. *J Pediatr* 99:223–227.
- Russell-Eggitt IM, Blake KD, Taylor DSI, Wyse RKH. 1990. The eye in the CHARGE association. *Br J Ophthalmol* 74:421–426.
- Vissers LE, van Ravenswaaij CM, Admiraal R, Hurst JA, de Vries BB, Janssen IM, van der Vliet WA, Huys EH, de Jong PJ, Hamel BC, Schoenmakers EF, Brunner HG, Veltman JA, van Kessel AG. 2004. Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. *Nat Genet* 36:955–957.
- World Health Organization. 1992. International statistical classification of diseases and related problems. 10th revision. Vol. 1. Geneva, Switzerland: World Health Organization.
- Zentner GE, Layman WS, Martin DM, Scacheri PC. 2010. Molecular and phenotypic aspects of CHD7 mutation in CHARGE syndrome. *Am J Med Genet Part A* 152A:674–686.

