

Frequency of holoprosencephaly in Chile ECLAMC: 41 años de vigilancia de la holoprosencefalia en Chile. Período 1972-2012

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© 2015, Sociedad Medica de Santiago. All rights reserved. Background: Holoprosencephaly is a structural anomaly of the brain that consists in a defect of the prosencephalon development that leads to face and neurological defects of variable intensity. Aim: To estimate holoprosencephaly prevalence at birth. Patients and Methods: All cases of holoprosencephaly, born alive or stillbirths, registered in the 15 Chilean Hospitals of the Latin American Collaborative Study of Congenital Malformations (ECLAMC) between 1972 and 2012, were studied. Craniofacial and other anomalies found in newborns affected by holoprosencephaly are described. Results: Fifty five cases of holoprosencephaly (58% males) were found among the 798.222 registered births (rendering a prevalence at birth of 0.69 per 10.000 newborns). The most common cranial defect was medial cleft lip with cleft palate (27.3%), bilateral cleft lip (11%) or both (38.2%), cyclopia (14%), single nostril (10.9%) and proboscis (9.1%). Eleven pe