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Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. [Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome](#).
ORPHA:52055

Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome is a developmental anomalies syndrome characterized by coloboma of the iris and optic nerve, facial dysmorphism (high forehead, microretrognathia, low-set ears), intellectual deficit, agenesis of the corpus callosum (ACC), sensorineural hearing loss, skeletal anomalies and short stature.