Open Peer Review on Qeios

Omphalocele syndrome, Shprintzen-Goldberg type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Omphalocele syndrome, Shprintzen-Goldberg type. ORPHA:3164

ShprintzenGoldberg omphalocele syndrome is a very rare inherited malformation syndrome characterized by omphalocele, scoliosis, mild dysmorphic features (downslanted palpebral fissures, s-shaped eyelids and thin upper lip), laryngeal and pharyngeal hypoplasia and learning disabilities.