

Meckel syndrome affects males and females in equal numbers. More than 200 cases have been reported in the medical literature. The incidence of Meckel syndrome is estimated in various areas around the world to be 1 in 13,250 to 1 in 140,000 live births. The disorder is more common in the Finnish population due to a founder effect, with an incidence of 1 in 9000 and 1 in 3,000 people of Belgian ancestry. However, Gujarati Indians have a prevalence of 1 in 1,300. It often occurs in the context of consanguineous unions. A diagnosis of Meckel syndrome is often made on ultrasound during pregnancy or at birth through clinical evaluation. Molecular genetic testing can be used to confirm the diagnosis and guide genetic counseling. Prenatal diagnosis is available through ultrasonography as early as 14 weeks, which can detect certain abnormalities (e.g., encephalocele, polydactyly, cystic kidneys and oligohydramnios). Magnetic resonance imaging (MRI) may be used in conjunction with ultrasonography. Chromosomal analysis may be performed to rule out trisomy 13. Smith Lemli- Optiz syndrome may be excluded by biochemical testing.