Fryns syndrome

Genetics

-No gene known

-AR

Clinical findings/Dysmorphic features

-Most common autosomal recessive syndrome associated with congenital diaphragmatic hernia

-Diaphragmatic defects (diaphragmatic hernia, hypoplasia or agenesis); pulmonary hypoplasia; distal digital hypoplasia (nails, terminal phalanges); genitourinary malformations

-Agenesis of the Corpus Callosum; optic and olfactory tract hypoplasia; encephalocele

-Facial: coarse facies; hypertelorism; broad/flat nasal bridge; thick nasal tip; long philtrum; low-set/poorly formed ears; tented upper lip; macrostomia (wide mouth); micrognathia

Etiology

-7 in 100,000 live births in a French population

Pathogenesis

-Not known

Genetic testing/diagnosis

-Based on clinical findings; several different chromosome aberrations have been described in individuals who have previously received a diagnosis of Fryns syndrome

Others

-Majority are stillborn or die in early neonatal period, 14% survive longer