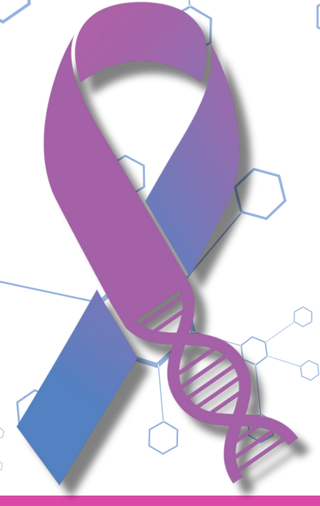
EMANUEL SYNDROME



WHAT SHOULD WE BE LOOKING FOR?

Common Findings in Emanuel Syndrome

Neurology: hypotonia, seizures, structural abnormalities including microcephaly, atrophy, ventriculomegaly, hydrocephalus, absent or hypoplastic corpus callosum, white matter abnormalities, Dandy-Walker malformation, Chiari malformation, tethered spinal cord (sacral dimple is also a common finding, in rare instances associated with a tethered spinal cord).

Cardiovascular: Atrial septal defects, ventricular septal defects, patent ductus arteriosus, and other, less common heart malformations.

Orthopedic: hip dysplasia, hip dislocation, scoliosis, kyphosis, foot malformations, ankle instability, joint contractures, torticollis.

Vision: Myopia, strabismus, astigmatism, ptosis, hyperopia, and other vision differences.

Hearing: varying degrees from mild to profound hearing loss is common. Hearing loss can be conductive or sensorineural.

Dentition: Delays in eruption of primary teeth, misaligned teeth, crowding, absent adult teeth, enamel defects

Craniofacial: ear pits and tags, changes in ear shape, size and position, cleft palate (overt or submucous), Pierre Robin sequence, small jaw, glossoptosis, laryngomalacia and tracheomalacia.

Gastrointestinal: inguinal hernia, diaphragmatic hernia, intestinal malrotation, imperforate anus.

Renal: small or single kidneys, vesicoureteral reflux, and other kidney differences.

Genital and anal: small penis, hypospadias (urethral opening on underside of penis), undescended testes, imperforate anus, Hirschprung's disease.

Gastrointestinal: drooling, aspiration, constipation, failure to thrive, choking and swallowing issues, gastroesophageal reflux.

Respiratory: recurrent chest infection, aspiration, apnea, tracheomalacia, laryngomalacia.

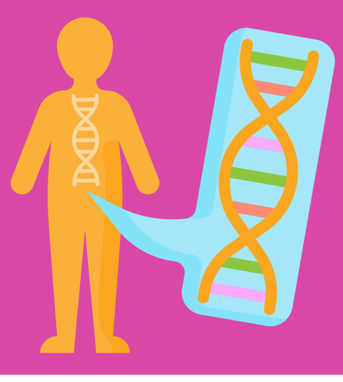
Musculoskeletal: ankle instability, hip subluxation or dislocation, scoliosis and kyphosis, joint contractures, torticollis.

Immunity: Recurrent infections of the ear, lungs (pneumonia), sinuses, urinary tract, recurrent oral candidiasis, low immunoglobulins.

Development: Speech and language therapy referral, physical and occupational therapy referral, monitor height and weight, and feeding assessment. Referrals should be made to early intervention or infant development specialist.

Surgical considerations: To assess for issues with sedation and difficult intubation, responses to anesthetic agents.

Genetic counseling: for parents or siblings to investigate carrier status of the t(11;22).



This is not a complete picture of ES and not all people exhibit every symptom.

Developed by: Chromosome 22 Central
1129 Carolina Gardens Ave., Fuquay-Varina, North Carolina, 27526 USA

EMAIL: c22central@gmail.com | tel: 919-762-7979

www.EmanuelSyndrome.org