Edwards Syndrome

Edwards syndrome is also known as trisomy 18, as this typically results from nondisjunction of chromosome 18, leading to three chromosomal copies. Patients display intellectual disability, along with rocker bottom feet. VSD is associated with Edwards syndrome, and children with this disorder display clenched hands with overlapping fingers. Facial features include low set ears, micrognathia and a prominent occiput. GI abnormalities associated with this syndrome include omphalocele and Meckel's diverticulum. Commonly, patients have a horseshoe kidney, and can develop malrotation of the gut.



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Pathophysiology

Trisomy 18
Tricycle (18) ID at Voting booth
Edwards syndrome is caused by the presence of a complete or partial extra copy of chromosome 18.

Signs and Symptoms

Intellectual Disability (Mental Retardation)
Tar Covered Book
Children with Edwards syndrome display significantly impaired cognitive functioning and deficits in two or more adaptive behaviors.

Rocker Bottom Feet
Rocking-chair with Rocker Bottom Feet
Rocker bottom feet are characterized by a prominent calcaneus and a rounded bottom of the foot, resembling the bottom of a rocking chair.

Ventricular Septal Defect (VSD)
V-ventricle Scepter heart defect
Ventricular septal defect is a congenital heart defect that enables blood to flow between the left and right ventricle. VSDs are common in children with Edwards syndrome. Other congenital heart defects may occur like PDA, ASD, and ToF. However, VSD is most common.

Clenched Hands
Clenched fist
Persistently flexed finger joints are a common physical finding in Edwards syndrome.

Overlapping Fingers
Overlapping Fingers
Patients with Edwards syndrome often exhibit overlapping fingers. Hands can present with overlapping digits, in which the second and fifth fingers sit atop the third and fourth fingers, respectively.

Low-Set Ears
Low set ears
Low-set ears are a common physical finding in Edwards syndrome.

Micrognathia
Small-jaw with Micro-gnats
Micrognathia is a small jaw, which is a common physical finding in Edwards syndrome.

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Prominent Occiput

Prominent Octopus

An enlarged protruding occiput is a common physical finding in Edwards syndrome.

Omphalocele

O-full-of-eels

Omphalocele is associated with Edwards syndrome. This is an abdominal wall defect leading to the intestines, liver and sometimes other organs extruding out of the abdomen, while being contained in a peritoneal sac. This occurs at the midline and the outpouching occurs at the navel.

Meckel's Diverticulum

Meckel-mouse Diver

Meckel's diverticulum is a common gastrointestinal defect associated with Edwards syndrome. This is a vestigial remnant of the vitelline duct, and is described as a slight bulge in the small intestine.

Horseshoe Kidney

Horseshoe Kidneys

Up to 20% of patients with Edwards syndrome have genitourinal defects, such as horseshoe kidney. Other defects include hydronephrosis and cystic kidneys.

Malrotation of Intestines

Mallet-rotating Intestines

Malrotation of the intestine is a congenital anatomical anomaly that results from an abnormal rotation of the gut as it returns to the abdominal cavity during embryogenesis. Occurance of this anomaly is believed to be due to hereditary factors, or an underlying genetic disorder, such as Edwards syndrome.

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