

Adams-Oliver syndrome (AOS) is an extremely rare inherited disorder characterized by defects of the scalp and abnormalities of the fingers, toes, arms, and/or legs. The physical abnormalities associated with this disorder vary greatly among affected individuals. Some cases may be very mild while others may be severe. In infants with Adams-Oliver syndrome, scalp defects are present at birth (congenital) and may include one or multiple hairless scarred areas that may have abnormally wide (dilated) blood vessels directly under the affected skin. In severe cases, an underlying defect of the bones of the skull may also be present. In addition, infants with this disorder typically have malformations of the hands, arms, feet, and/or legs. These range from abnormally short (hypoplastic) fingers and toes to absent hands and/or lower legs. In some cases, additional abnormalities may also be present. Most cases of AOS appear to follow autosomal dominant inheritance but autosomal recessive inheritance has also been reported. Adams-Oliver syndrome is an extremely rare disorder that appears to affect males and females in equal numbers. More than 125 affected individuals have been reported in the medical literature. The major physical features of Adams-Oliver syndrome (i.e., scalp defects and limb abnormalities) are apparent at birth (congenital). The diagnosis of Adams-Oliver syndrome may be suspected at birth based upon the identification of characteristic scalp and skull defects occurring in association with malformations of the fingers, toes, hands, and/or feet. The diagnosis may be confirmed by a thorough clinical evaluation, a detailed patient history, and a variety of specialized tests, such as advanced imaging techniques. In some cases, it may be possible to detect defects of the skull and/or limbs before birth (prenatally) through the use of advanced imaging techniques such as ultrasound. During fetal ultrasonography, reflected sound waves are used to create an image of the developing fetus.