CAHMass

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Categories: Laboratory Infrastructure

Short Description:

Congenital adrenal hyperplasia (CAH) refers to a group of autosomal recessive disorders that impair cortisol biosynthesis. CAH represents a continuous phenotypic spectrum with over 95% of all cases caused by 21hydroxylase deficiency. CAH owing to 21hydroxylase deficiency is the most common cause of genital ambiguity in the newborn and is present in about 1 in 15000 live births worldwide

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Congenital adrenal hyperplasia (CAH) refers to a group of autosomal recessive disorders that impair cortisol biosynthesis. CAH represents a continuous phenotypic spectrum with over 95% of all cases caused by 21hydroxylase deficiency. CAH owing to 21hydroxylase deficiency is the most common cause of genital ambiguity in the newborn and is present in about 1 in 15000 live births worldwide Features Liquid Chromatography Tandem mass spectrometry (LCMSMS) techniques allows for the simultaneous specific determination of 17OHP and other steroids related with CAH such as androstenedione cortisol 11deoxycortisol and 21deoxycortisol. Application of this technology to the determination of steroids in newborn blood spots significantly enhances the correct identification of patients with CAH and reduces the number of falsepositive screening results when implemented as a secondtier analysis performed prior to reporting of initial newborn screen results.