**Introduction**

Coffin Siris syndrome (CSS) is a congenital disease characterized by a coarse facies and hypoplasia or aplasia of distal phalanx or nail (typically the 5th finger) which affects neurological development and the immune system. Various mutations (mainly de novo) have been implicated, affecting BAF or PBAF complexes and their subunits implicated in chromatin remodeling.

**Case Report**

**Prenatal**

We report a case of prenatal diagnosis of CSS. The mother had no prior medical history. The first ultrasound showed no abnormality. At 18 weeks’ gestation, ultrasound showed hydrocephalus and macrocephaly (Fig. 1), a left diaphragmatic hernia, severe IUUG, single umbilical artery (Fig 2) and a ventricular septal defect (Fig 3). She requested a medical termination which was accepted according to French law.

**Postnatal**

- The autopsy showed an aplasia of the distal phalanx (5th finger, Fig 6), a coarse facies and malformations were confirmed (Fig 4-6).
- Genetic testing found a de novo ARID1A mutation which is responsible for CSS.

**Discussion**

Our case highlights prenatal signs that could lead to evoking the diagnosis:

- IUUG is the most frequent sign described prenatally.
- Ventriculomegaly had never been described in CSS.

Cardiac malformations or cleft palate have been reported in infants with CSS. Their prenatal description remains extremely rare. In one recent case report (Sweeney et al., Cold Spring Harb Mol Case Stud; 2018), ultrasound showed left diaphragm hernia with IUUG, aortic arch hypoplasia, small left sided cardiac structures and a ventricular septal defect. The karyotype was normal, and the diagnosis of CSS was based on clinical findings and whole genome sequencing after birth (ARID1B mutation). To our knowledge, our case and this one are the most severe cases described so far.

As prenatal presentation of CSS is rare, especially when severe, our case helps to understand this pathology. Yet, associated malformations are non-specific. Therefore, the possibility of prenatal diagnosis appears limited in the absence of an index case.