

EP10.27 Polyhydramnios as a prenatal clue to congenital myotonic dystrophy: case report and review of 28 pregnancies

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Objectives

Congenital myotonic dystrophy (CDM) is a severe neonatal disorder with nonspecific prenatal findings. We present a case and summarize prenatal features and outcomes.

Methods

We report a case of CDM diagnosed postnatally after polyhydramnios and hydrops. A literature review identified pregnancies with CDM and reported prenatal findings. Outcomes were analyzed descriptively

Results

Twenty-eight pregnancies (27 published, 1 institutional) were analyzed. Our case involved a 33-year-old presented at 33 weeks with severe polyhydramnios (AFI 55 cm) requiring amnioreduction, followed by hydrops and delivery at 34 weeks. The neonate had hypotonia and respiratory failure; CDM was confirmed, with subsequent maternal diagnosis. Polyhydramnios was the most common finding (82%), usually in the third trimester (~30 weeks). Other findings included ventriculomegaly (25%), talipes (18%), pleural effusion (18%), and hydrops (14%). Prenatal diagnosis occurred in 21% of cases. Preterm delivery occurred in 50% and cesarean delivery in 82%. Hypotonia occurred in 93%, respiratory support in 75%, and mortality in 29%.

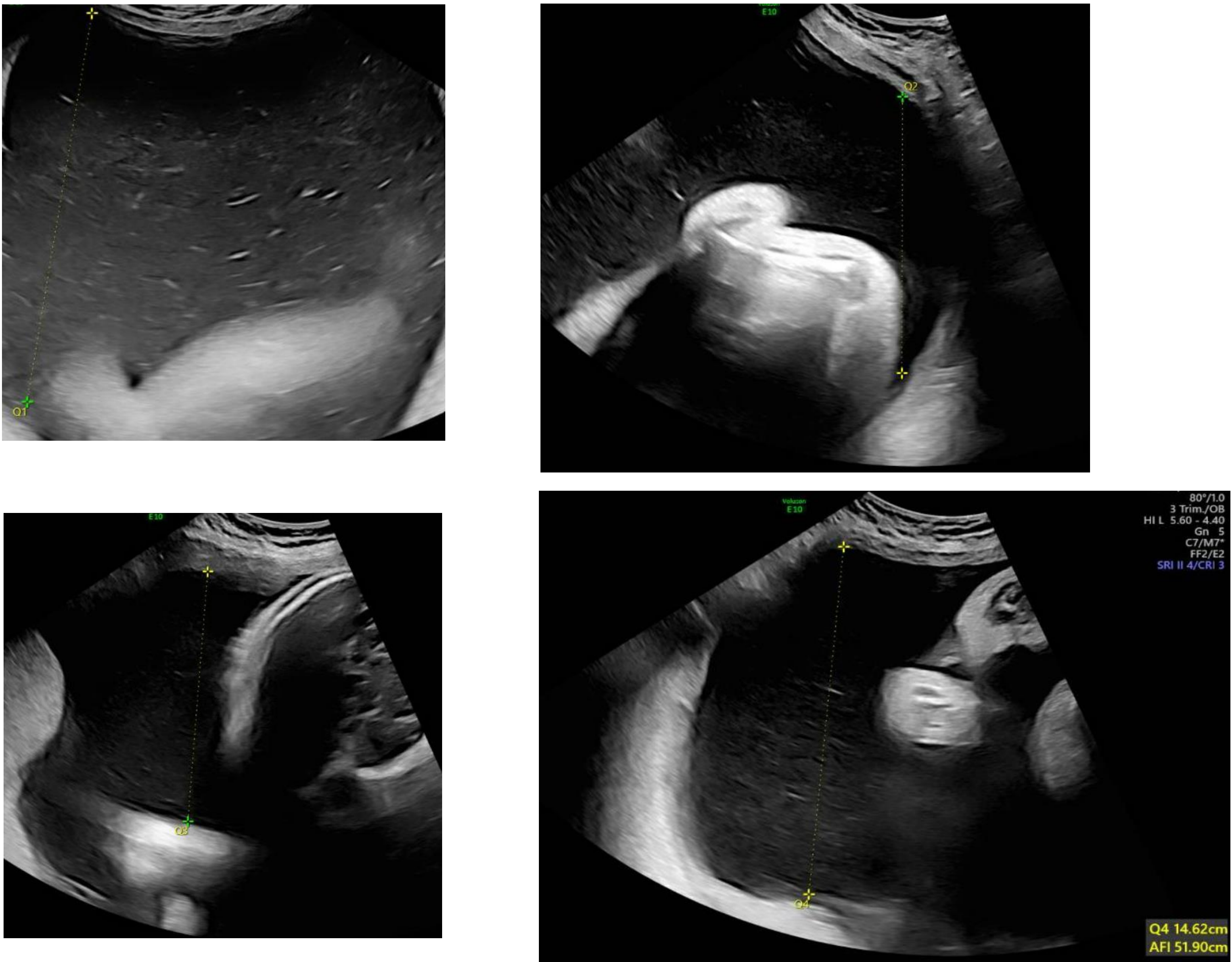
Table

TABLE:
Key Prenatal Ultrasound Findings and Neonatal Outcomes in Pregnancies Affected by Congenital Myotonic Dystrophy (n = 28)

Variable	Value
Polyhydramnios	23/28 (82.1%)
Ventriculomegaly	7/28 (25.0%)
Talipes	5/28 (17.9%)
Pleural effusion	5/28 (17.9%)
Hydrops fetalis	4/28 (14.3%)
Prenatal diagnosis of DM1	6/28 (21.4%)
Preterm delivery	14/28 (50.0%)
Cesarean delivery	23/28 (82.1%)

Hypotonia	26/28 (92.9%)
Respiratory support	21/28 (75.0%)
Neonatal mortality	8/28 (28.6%)

Figure



Conclusion

Unexplained polyhydramnios, especially with decreased fetal movement or hydrops, should prompt consideration of CDM even with unknown maternal history.