Editorial

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The Obstetrician-Gynecologist and Rare Diseases

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Assuming that every pregnancy is a precious pregnancy, sometimes the fruit of a long period of hypo-fertility is an expensive therapeutic arsenal.

These pathologies are rare from the professional's point of view; but in the parent's eyes throughout their lives.

The practitioner is thus faced with cases of rare diseases where he must ensure a maternal care and a prognosis fetal and of course to be able to answer several questions:

1. Viable fetus or not?
2. Malformation type?
3. The fetal and postnatal prognosis?
4. And also the maternal prognosis?

For all these reasons, all the efforts of all the practitioners of the world must be gathered by publishing their experiences and enrich the medical bibliography in order to communicate these rare pathologies, facilitate their diagnosis and try to develop a consensual management .

To be pragmatic; I cite some cases [1-5] that I met in my professional life:

Case 1: Successful pregnancy by IVF in a patient with congenital cervical atresia [1].

Take home message: The case suggests that successful pregnancy in patients with congenital cervical atresia but functional uterus could be achieved by ART, no matter whether cervical reconstruction could be achieved or not.

Case 2: Twin pregnancy with both complete hydatidiform mole and co-existent alive fetus [2].

Take home message: We carefully suggest that in a CHMCF with a normal karyotype and no gross abnormalities on sonography, pregnancy may be continued as long as maternal complications are absent or, if present, controllable.

Case 3: Facial teratoma in the Newborn [3].

Take home message: The diagnosis of these rare cases of teratomas is possible in antenatal by ultrasound. The prognosis depends essentially on the histological nature, the site and size of the tumor. Generally favorable after surgical treatment and multidisciplinary care; but can be severe with neonatal death in other cases.

Case 4: Arthrogryposis multiplex congenital [4].

Take home message: The antenatal ultrasound diagnosis of arthrogryposis is often possible when the examination is done by an expert. The finding of an oligohydramnios should indicate a more detailed ultra-sonographic examination in search of other anomalies.

Recent research in molecular genetics and immunohistochemistry seems to be useful in clarifying certain etiologies.

Case 5: Antenatal bilateral renal vein thrombosis with combined protein S and C deficiency [5].

Take home message: Neonatal RVT continues to pose significant challenges for neonatologists. Recent data suggest that infants with heritable thrombophilia are more exposed to in utero and/or bilateral involvement and recurrent thromboembolic disease.

All these cases among many others; encourage health professionals to multiply our efforts in order to be able to manage properly these pathologies qualified as exceptional (but which do not cease to increase their incidences!) and meet the wishes and expectations of our patients.

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