detectable only by the use of molecular cytogenetics, the most commonly used in medical practice being Fluorescent In Situ Hybridization FISH, which locates a specific sequence using a complementary probe of fluorescently-labeled DNA. In most cases, de novo deletions are involved; not transmitted by the parents but linked to a meiotic accident. The aim is to study the clinical and evolutionary features of microdeletional syndromes diagnosed in the neonatal period in the neonatology department of Farhat Hached hospital in Sousse, Tunisia.

Methods: A retrospective, descriptive study of all microdeletional syndromes diagnosed in the neonatology department of Sousse, over a 10-year period (January 2014-December 2023). Diagnostic orientation was essentially clinical, radiological and biological. The molecular genetic study was carried out in the Cytogenetics Department of the same hospital, based on karyotyping with FISH and molecular analysis using multiplex PCR.

Results: We collected 24 cases of microdeletional syndromes, equally distributed between males and females. Diagnosis was made antenatally in 3 cases. Prematurity was found in 7 cases. Signs suggesting a karyotype and molecular study were facial dysmorphia (15 cases), congenital heart disease (13 cases), unexplained hypotrophy (11 cases) and neurological distress (10 cases). The various microdeletional syndromes diagnosed by the FISH technique were, in order of frequency : DiGeorge syndrome (13 cases), Prader Willis syndrome (8 cases), Williams and Beuren syndrome (1 case), 1p36 syndrome (1 case) and cat's cry syndrome (1 case). The molecular alterations observed enabled us to establish a genotype/phenotype correlation between the genetic abnormality and the clinical presentation of our patients.

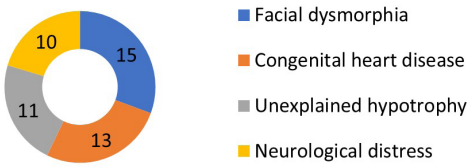

Fig 1. Clinical symptomatology

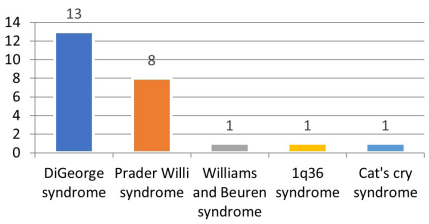

Fig 2. The microdeletional syndromes diagnosed

Conclusion: The contribution of molecular cytogenetics (FISH) is crucial for the investigation of microdeletional syndromes, with the aim of confirming the diagnosis, genetic counseling and management, which is usually multidisciplinary. At the end of this work, we have determined the optimal molecular diagnostic strategy for all microdeletional syndromes that can be diagnosed in the neonatal period, while focusing on the role of genetic counselling and prenatal diagnosis.

Keywords: Microdeletional syndrome, FISH, molecular cytogenetics, genetic counseling

PP-035 Hydrops fetalis: etiologies, management and outcome

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Objective: Hydrops fetalis (HF) is defined as excessive accumulation of amniotic fluid in the extravascular fetal compartment and at least two serous cavities. It manifests as subcutaneous edema, pleural or pericardial effusion and ascites. Etiologies are classified as immunological and non-immunological.

The objective of this study is to identify the clinical, etiological, therapeutic and evolutionary characteristics of HF in the neonatal period.

Methods: It is a descriptive and retrospective study, having included all newborns (NB) hospitalized in the neonatal intensive care unit of Farhat Hached Hospital of Sousse for management of HF, over a period of 7 years (January 2017- December 2023).

Results: The study included 20 NB. Sex ratio (F/M) = 2/3. Mean gestational age was 37 weeks' gestation (WG). HF was diagnosed during pregnancy in 17 women at a mean term of 28 WG. Hydramnios was associated in 12 cases and pulmonary hypoplasia in 6 cases. The pregnancy was complicated by gestational diabetes in 6 cases. All NB required resuscitation with intubation in the delivery room. Clinical signs were dominated by subcutaneous oedema in 18 cases, large ascites in 12 cases, pleural effusion in 10 cases, hepatosplenomegaly in 9 cases and pallor in 5 cases. Other signs were facial dysmorphia in 6 cases, heart murmur in 2 cases and tachycardia in 1 case. Cardiac origin was the predominant etiology, with one case of atrial flutter, one case of Ebstein's disease,

one case of transposition of great arteries and 2 cases of coarctation of the aorta. Intra-thoracic malformations were found in 3 patients (diaphragmatic hernia (2 cases) and digestive duplicity (1 case)). Anemia was the etiology of hydrops in 5 cases, of infectious (2 cases) or immunological (3 cases) origin, two of which received an in utero transfusion. Overload disease was found in 2 cases: mucopolysaccharidosis in 1 case, sialidosis type 2 in 1 case. One patient had congenital hypothyroidism and another a cystic lymphangioma. The etiology remained undetermined in 3 cases. Therapeutically, mechanical ventilation was used in all NB with surfactant instillation in 6 cases. Effusion puncture was performed in 8 cases. Exchange transfusion was used in cases of fetomaternal alloimmunization. Only 3 cases (atrial flutter after electrical and drug-induced cardioversion, rhesus alloimmunization and cystic lymphangioma) had a favorable outcome.

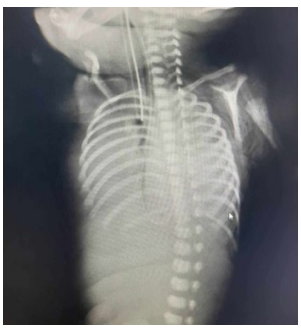

Fig 1. Chest Xray : Diaphragmatic hernia

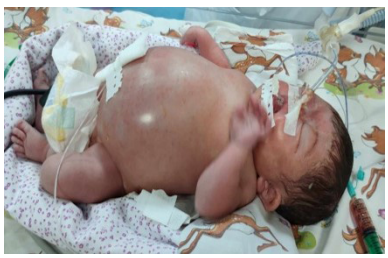

Fig 2. Subcutaneous oedema with large ascites and tachycardia

Conclusion: HF is a severe life-threatening neonatal disorder. The incidence of immunological etiologies has declined considerably since the prevention of Rhesus alloimmunization by anti-D immunoglobulin prophylaxis.

Keywords: Hydrops fetalis, prenatal diagnosis, non-immune fetal edema, congenital heart disease

PP-036 The impact of the covid-19 pandemic on the prevalence of intrauterine fetal death in philippine general hospital

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Objective: This study aims to determine the profile of cases of intrauterine fetal death among pregnant patients who were admitted in the Philippine General Hospital during the COVID-19 pandemic from March 2020 to December 2021.

Methods: This retrospective cross-sectional study involved all COVID-19-positive and COVID-19-negative pregnant women who delivered stillbirths during the pandemic period (March 2020 – December 2021) at the UP-PGH. Maternal and fetal factors of each pregnancy were analyzed.

Results: From the 3199 deliveries, there were a total of 68 stillbirth cases during the COVID-19 pandemic in 2020-2021. The prevalence rate during the pandemic was higher, at 2.1% as compared to the pre-pandemic rate which was 1.1%. Sixty-four percent belong to the 19 to 34-year-old age group, 13.2% were obese, 66.2% were multiparous, 33.8% high school as the highest educational attainment, 75% were single and 41.5% had at least 1-3 prenatal visits.

There was not enough evidence to show a significant difference in the demographic and clinical characteristics between those with positive and negative COVID-19 disease. Fifty-one cases (75%) were COVID-19 negative and 17 (25%) were COVID-19 positive. There was not enough evidence to show that any additional cause of IUFD was associated with COVID-19 disease, based on the demographic and clinical characteristics.

Conclusion: There was no statistically significant difference in the prevalence of stillbirths pre-pandemic and the year during the COVID-19 pandemic in our institution. Moreover, the COVID-19 disease did not have a direct impact on the cases of stillbirths as evidence did not show that any additional cause of the IUFD was linked to the COVID-19 disease. This can be interpreted as COVID-19 disease was not significantly associated with the negative effect on pregnant women and their fetuses but rather that the pandemic's effect on government policies and restrictions as well as admission protocols at our institution were to be considered as contributing factors affecting the stillbirth cases during the pandemic.