20th World Congress in Fetal Medicine

# Precision medicine in perinatal cardiology: single center experience

Muñoz H, Enriquez G, Cortez F, Germain A, Triantafilo J, De la Jara C, Perez AL, Enriquez R, Robert JA, Palermo M
Universidad de Chile and Clinica las Condes, Santiago, Chile

## Objective

Our objective is to describe the cases of congenital heart disease (CHD) associated with single gene diseases or deletions (SGD), precision medicine.

## Methods

We design a descriptive retrospective study. We reviewed the neonatology and Rare disease Center database of Clínica las Condes between January 2015 and April 2023. Single gene disease was identified using MLPA (Multiplex Ligation-dependent Probe Amplification), gene panel or Exome analysis, from samples of amniotic fluid, or neonatal Blood, obtained during perinatal period. In each of the cases, the type of CHD and the SGD, molecular biology technique used, and the perinatal results are described.

## Results

During the period 2015 to 2023, 12 cases of CHD associated with SGD, 58,3 % (7) identified during the prenatal period and 41,7 % (5) in the postnatal period. Since 2022, all cases were diagnosed in utero and the most widely used molecular biology test was exome. The most common malformations were Hypertrophic Myocardiopathy (4) and Double outlet Right Ventricle (3). 58,3 % (7) were born alive, 1 intrauterine death and 4 terminations. Of those born alive, 5 of 7 died during the first year of life. 83% had poor perinatal outcomes.

## Conclusion

Precision Medicine in perinatal cardiology is possible. Until 2021, without pregnancy terminations, 16.1% of children with CHD, born in our center had SGD (9823 births, 137 CHD newborn, 30 major CHD, and 5 with SGD. By incorporating prenatal diagnosis and pregnancy termination in fetuses carrying heart disease with single gene diseases or deletions, the number of patients doubled in one year. The prognosis of children with CHD associated with SGD is poor and prenatal diagnosis makes it possible pregnancy termination and, in the future, gene therapy.