

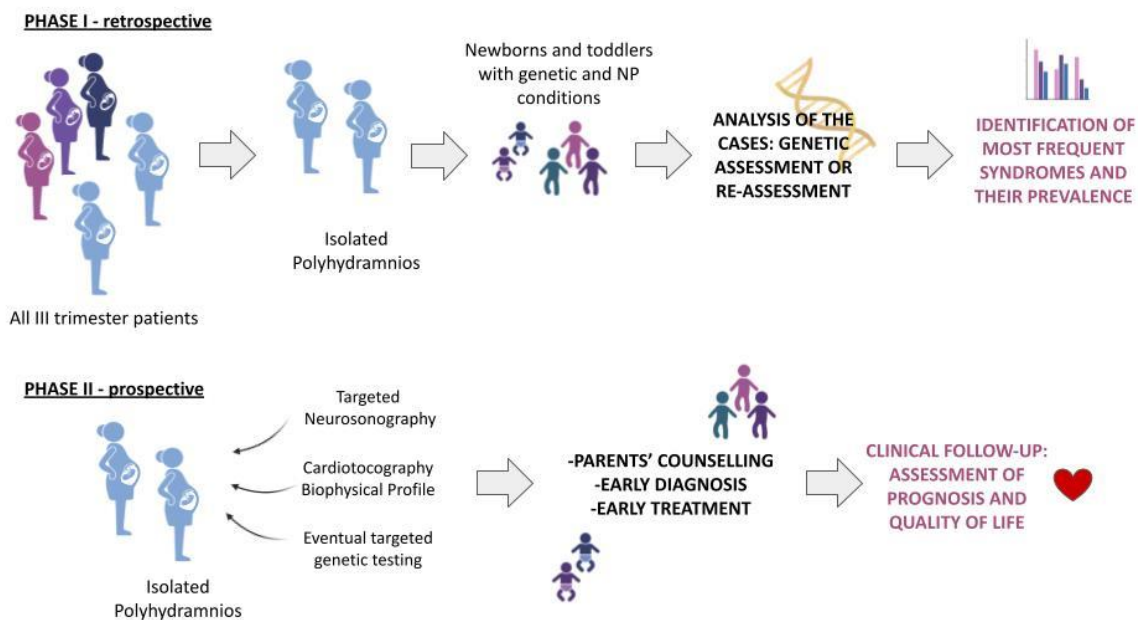
**Project title:** Idiopathic polyhydramnios: an understated indicator of genetic and neuropsychiatric conditions?

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**In collaboration with:**

- Prenatal Diagnosis Unit, Obstetrics and Gynecology, IRCCS Policlinico San Matteo
- Medical Genetics Unit, IRCCS Policlinico San Matteo
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**Background:** Polyhydramnios, i.e. an abnormally high quantity of amniotic fluid, is known to be associated with maternal diseases, placental dysfunction and fetal neurological or structural anomalies, such as interruptions of the gastrointestinal tract or renal problems. However, when no obvious maternal, placental or fetal abnormality is found, polyhydramnios could be the leading symptom of more subtle fetal conditions. Neurodevelopmental disorders, oro-buccal apraxias, dyspraxias can be not immediately evident even in the neonatal period, leading to underdiagnosed genetic and neuropsychiatric conditions and missed opportunities for early treatment.



**Scientific hypothesis and aims:**

We propose idiopathic polyhydramnios as potential fetal marker for genetic and neuropsychiatric conditions in the newborn and in the toddler. Moreover, we hypothesise that an early evaluation of isolated polyhydramnios would allow early treatment and benefit children and families.

By exploiting a large cohort of women with idiopathic polyhydramnios diagnosed in the 3rd trimester of pregnancy, we plan to:

- 1) retrospectively identify the children who underwent genetic and/or neuropsychiatric evaluation for neurodevelopmental and deglutition concerns
- 2) describe the phenotypes of the babies and propose genetic analysis when appropriate
- 3) prospectively follow-up pregnant patients with isolated polyhydramnios, performing targeted neurosonography. Biophysical profile and cardiotocography will be performed for the assessment of fetal movements. Prenatal diagnosis and genetic counselling will be carried out when needed.
- 4) Refer the babies with anamnesis of idiopathic polyhydramnios to an early neuropsychiatric evaluation, in order to provide early diagnosis and treatment when underlying conditions are present
- 5) assess clinically the children over the years and evaluate if any change of prognosis and quality of life is significant.

**Techniques:** Fetal ultrasound scanning, Neurosonography, Cardiotocography, Array-CGH, NGS panels.