# CALL FOR COLLABORATIONS FROM ACADEMIA AND INDUSTRY

- **Title of the study:** RARB Natural History Study

- **Targeted gene(s)/phenotype/disorder under study:** RARB (OMIM: 615524)

- **Summary:**

*RARB*-related disorder, also known as MCOPS12, is caused by pathogenic variants in the *RARB* gene. *RARB* codes for retinoic acid receptor beta, a transcription factor belonging to the family of nuclear receptors. This condition is characterized by developmental eye defects (including microphthalmia and coloboma), other congenital anomalies (including diaphragmatic hernia and heart defects), and global developmental delay with dystonia. Although most affected individuals display severe developmental delay and motor impairment, some show minimal neurodevelopmental involvement. There is great allelic and phenotypic heterogeneity described in this rare and understudied disorder.
As most reported cases are children, we initiated a natural history study of MCOPS12 to better characterize its clinical course across the lifespan. Individuals of any age, from any country, carrying pathogenic or likely pathogenic variants in *RARB* are eligible to participate. The study entails annual participant and physician questionnaires, as well as uploading brain MRIs and participant videos. Fetal and deceased cases are also eligible to participate, with modifications to the study. Our study will lead to the establishment of a framework for the development of clinical trials against MCOPS12.

- **Coordinating clinician**:

Jacques Michaud – principal investigator
Valerie Chu – genetic counsellor and study coordinator

- **Institution (dept, hospital, City)**: CHU Sainte-Justine, Montréal, Canada
- **Contact email**:

Jacques.michaud.med@ssss.gouv.qc.ca
valerie.chu.hsj@ssss.gouv.qc.ca