The challenge of fetal phenotyping

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Objective
To present state-of-the-art solutions to characterise prenatal findings (fetal phenotypes) and applications in Fetal Medicine (phenotype/genotype correlations, prenatal/postnatal correlations).

Methods
A shared representation of fetal phenotypes is critical in Fetal Medicine to investigate the pathological processes involved, to establish the postnatal prognosis and to drive prenatal treatment options. The correlations between a precise and standardised description of fetal phenotypes with genetic investigations (WES, WGS) and with postnatal phenotypes is key to a global advance in imaging, genetics and prenatal counselling based on evidence. Establishing these correlations impose a shared manner to description of fetal findings, especially from imaging investigations.

Results
The human phenotype ontology (HPO) has become a standard for the postnatal representation of phenotypes in clinical genetics. An international working group has addressed the extension of HPO to cover prenatal phenotypes. Additionally, a European initiative (France, Spain, Belgium, UK) is developing an extensive ultrasound image collection with fetal phenotypes.

Conclusion
We will present the current status of the fetal phenotype description and its applications in Fetal Medicine care, research and potential artificial intelligence outcomes.