A new Prenatal Diagnosis paradigm non- focused on Down syndrome

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Prenatal diagnosis of birth defects was initially focused on Down syndrome and neural tube defects. In many countries, an anomaly ultrasound scan is offered to pregnant women to screen for any kind of fetal structural defect, that account for a 3% prevalence at birth. Although genetic anomalies have a similar 3% prevalence, prenatal diagnosis programs have not been expanded similarly. A more comprehensive paradigm should be enforced, including information on other genetic causes than Down syndrome for intellectual disability and neurodevelopmental impairment, that may be divided in the 3 groups of genetic anomalies, chromosomal, subchromosomal and single-gene disorders. Ideally, cell-free DNA or invasive karyotyping (for chromosomal anomalies), invasive microarray analysis (for subchromosomal anomalies), and carrier screening for common recessive disorders (for single-gene disorders), would be offered and discussed with each pregnant woman.