Supplementary figure 2a. Secondary analysis: detailed workflow of the proposed algorithm.
Supplementary figure 2b. Secondary analysis: detailed workflow of the current algorithm

Patients with primary indications of invasive prenatal diagnosis

CVS or amniocentesis $n=130$

- Ultrasound abnormal / increased NT $n=73$
  - Cell culture $n=73$
    - Abnormal/Inconclusive results $n=20$
      - QF-PCR for common aneuploidies detection
        - Normal results $n=22$
          - aCGH for CNV detection
            - Normal results $n=39$
              - Confirmatory tests (e.g., MLPA, FISH) $n=5$
            - Abnormal results $n=42$
        - Those willing to pay for aCGH (41.8%) $n=53$
          - QF-PCR for common aneuploidies detection* $n=23$
            - Normal results $n=20$
              - aCGH for CNV detection
                - Normal results $n=42$
                - Abnormal results $n=3$
            - Abnormal/Inconclusive results $n=3$
          - DS screening positive / family history of chromosomal or genetic disorders $n=57$
            - Cell culture
              - Abnormal/Inconclusive results $n=3$

Karyotyping** $n=130$

- Reporting

$n=4$