1. NIPT: an overview

NIPT is a potential emerging field in genetic prenatal diagnosis and screening. Thus, NIPT relies on the identification of bidirectional traffic between the fetus and the mother through the placenta.

Bloodstream. The different epigenetic pattern is an example of an useful method to detect fetal markers. In addition, NIPT is performed with the aim to identify fetal genetic material in maternal bloodstream so as to detect genetic abnormalities.

Advantages

1. Dilution
2. Real
3. Counting
4. Measure

Disadvantages

Economic, ethical, social and legal issues should be considered due to the ease and absence of risk performing NIPT.

3. Down Syndrome

Down syndrome (DS) or trisomy 21 is the most common aneuploidy (1/800 live births) and the most common cause of severe mental delay. DS is compatible with life but it involves some characteristic clinical features (Table 1). The high mortality is due to heart disease which is very frequent. In addition, maternal age and previous family history are the main risk factors (Figure 5).

3.1. Clinical features

- Hypotonia
- Smaller nose and ears
- DENTUS LIUS
- Variable intellectual coefficient (45-70)
- Developmental delay
- Alzheimer susceptibility
- Leukemia susceptibility (at earlier ages)

Table 1. It is summarised the main clinical features in DS.

Figure 5. This graphic shows an exponential increase risk of carrying a DS baby as maternal age increases.

Owing to its high incidence, DS is the main reason why women undergo prenatal testing. The detection of DS prenatally involves screening and diagnostic testing. Currently, there are three screening algorithms designed with the aim to estimate a pregnancy-specific individual risk at carrying a DS fetus. These algorithms are based on the evaluation of three elements: maternal age, nuchal translucency (NT) and maternal serum biomarkers.

Figure 6 only shows the first trimester combined test which is recommended by ISPD (International Society for Prenatal Diagnosis). Depending on the estimated risk level various strategies can be followed (Figure 7).

4. Bioethical aspects

The development of NIPT has raised numerous ethical, social and legal issues so it is essential to evaluate the pros and cons (Figure 9). Some ethical concerns should be taken into account such as the right of “not to know” and the abortion.

5. Conclusions

- NIPT is performed with the aim to identify fetal genetic material in maternal bloodstream so as to detect genetic abnormalities. The absence of invasiveness allows to avoid additional risks caused by the procedure.
- NIPT can be applied for screening but not for definitive diagnosis.
- NIPT is a potential emerging field in genetic prenatal diagnosis and screening.
- NIPT is performed with the aim to identify fetal genetic material in maternal bloodstream so as to detect genetic abnormalities.
- The different epigenetic pattern is an example of a useful method to detect fetal markers.
- NIPT is performed with the aim to identify fetal genetic material in maternal bloodstream so as to detect genetic abnormalities.
- Economic, ethical, social and legal issues should be considered due to the ease and absence of risk performing NIPT.

6. References