NIPT: Genetic Conditions

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- **Trisomy 21 (Down Syndrome):** Occurs in about 1 in 450 live births in Ireland, characterised by an extra copy of chromosome 21, leading to developmental delays and distinct physical features. More info: NHS Down Syndrome.

- **Trisomy 18 (Edwards' Syndrome):** Occurs in approximately 1 in 2,000 pregnancies and is associated with severe developmental and health challenges due to an extra chromosome 18. Sadly babies with Edwards' Syndrome often have a high risk of death before or shortly after birth. More info: NHS Edwards' syndrome

- **Trisomy 13 (Patau's Syndrome):** Affects about 1 in every 4,000 births, characterised by an additional copy of chromosome 13. This severely disrupts normal development. Sadly this can often result in miscarriage, stillbirth or the baby dying shortly after birth. More info: NHS Patau's Syndrome.

- **Monosomy X (Turner Syndrome):** Affects 1 in every 2000 baby girls, characterised by the absence of one X chromosome, leading to various developmental issues. More info: NHS - Turner Syndrome.

- **XXY Syndrome (Klinefelter Syndrome):** When baby boy is born with extra X chromosome, occurs in about 1 in 1,000 male births, resulting from an extra X chromosome. Those with Klinefelter syndrome are still genetically male, and often will not realise they have this extra chromosome, but occasionally it can cause problems that may require treatment. More info: NHS Klinefelter Syndrome.

- **Triple X Syndrome:** Affects approximately 1 in 1,000 female births, usually resulting in taller stature and potential learning difficulties. More info: NHS - Triple X Syndrome.

- **XYY Syndrome (Jacob's Syndrome):** Occurs in about 1 in 1,000 male births and is often associated with taller than average height and potential learning difficulties. More info: NHS - XYY Syndrome.

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- **Triploidy:** Happens in approximately 1 in 10,000 live births and results from an extra set of chromosomes, often leading to miscarriage or stillbirth. More info: NIH Triploidy

- **22q11.2 Deletion Syndrome (DiGeorge Syndrome):** Occurs in about 1 in 4,000 live births, leading to heart defects, immune deficiencies, and developmental delays. More info: NHS DiGeorge Syndrome

- **1p36 Deletion Syndrome:** Affects around 1 in 5,000 live births, associated with developmental delays and distinct facial features. More info: NIH 1p36 Deletion Syndrome.

- **Angelman Syndrome:** Occurs in about 1 in 15,000 live births. A condition that affects the nervous system and causes severe physical and learning disabilities. More info: NHS Angelman Syndrome

- **Cri-du-chat Syndrome:** Happens in approximately 1 in 15,000 to 1 in 50,000 live births, leading to developmental delays and distinctive features. More info: NIH Cri-du-chat Syndrome.

- **Prader-Willi Syndrome:** Occurs in about 1 in 15,000 live births and causes a wide range of physical symptoms, learning difficulties and behavioural challengesis including hyperphagia (excessive eating), learning difficulties and weak, floppy muscles (hypotonia) . More info: NHS Prader-Willi Syndrome

- **Wolf-Hirschhorn Syndrome:** Affects approximately 1 in 50,000 live births, resulting from a deletion of part of chromosome 4 and associated with severe developmental delays and distinct facial features. More info: NIH - Wolf-Hirschhorn Syndrome.

Important Note

The prevalence rates of these conditions can vary based on multiple factors, including maternal age and genetic predisposition.