Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions

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Many babies with conditions associated with 22q11.2DS require urgent medical attention at birth or soon afterwards. **Early diagnosis** can help provide the best after-birth care, which is especially important for newborns who have critical heart defects, but can also help those with more subtle health issues avoid a lengthy search for a definitive answer.

This article provides prenatal guidance for: (1) families with no history of 22q11.2DS; (2) prospective parents who have 22q11.2DS themselves; and (3) unaffected couples who already have a child with 22q11.2DS. There is discussion about **methods of obtaining samples** for prenatal genetic screening and testing, as well as physical features that may be seen on **imaging**. There is explanation about what each **testing method** can and cannot detect, and the importance of **genetic counselling**.