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.