Twenty-four hour, Non-invasive, Neonatal Chromosome Analysis—Application in a Case of Mixed Gonadal Dysgenesis

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Abstract

***Introduction**: With the advent of interphase molecular fluorescent* **in-situ** *hybridisation (FISH), buccal mucosa can be used to provide an highly accurate assessment of those chromosomes most commonly causing abnormality in live-born children. **Clinical Picture**: A newborn child presented with ambiguous genitalia. The phallus-looking enlarged "clitoris" had a urethral opening at the ventral surface near the tip and the "labial" folds were completely fused. No definite gonads were palpable. Differential diagnostic possibilities included sex chromosome or a single gene abnormality such as congenital adrenal hyperplasia. Thus, one initial objective was to investigate the sex chromosomes. Buccal mucosa was used in conjunction with fluorescent molecular probes for the X and Y. This methodology enabled a firm diagnosis of a 45,X/46,XY mosiac to be made within 24 hours. Decisions could then be made concerning gender assignment. **Treatment and Outcome**: Intervention by means of reconstructive surgery of the external genitalia would be made available at a later date. **Conclusions**: The use of buccal mucosa is non-invasive, easy to obtain and, when combined with molecular techniques, is reliable and accurate. The clinical implication of this methodology is that it will be especially useful in gender assignment or when rapid decisions on live-saving surgery have to be made in cases of possible aneuploidy.*

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Key words: Buccal mucosa, FISH, Gonadal ambiquity, Probe, Mosaic, Sex chromosomes

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