

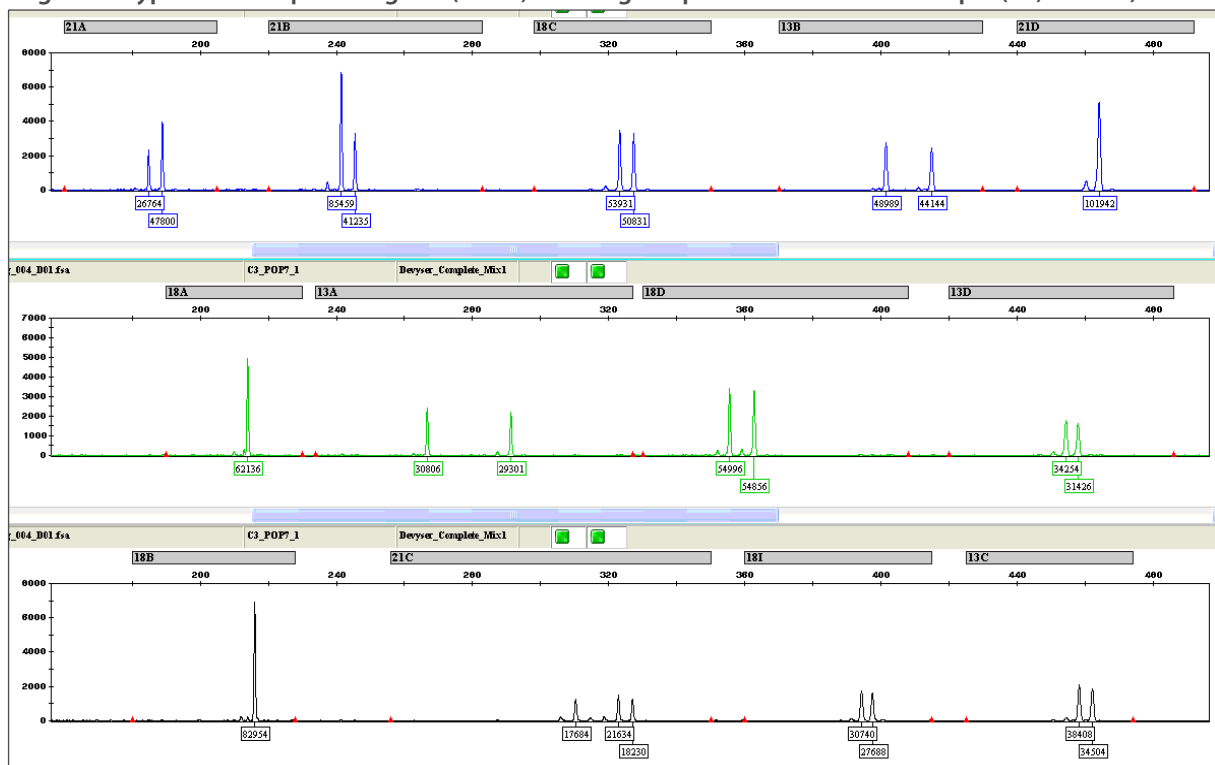


Devyser® Complete QF-PCR kit for Prenatal Diagnostics

Conclusions

- The Devyser® Complete kit shows 100% correlation to karyotyping results for the detection of common autosomal and sex chromosome aneuploidies.
- The Devyser® Complete kit shows 100% sensitivity (14/14 samples) for detection of the common autosomal and sex chromosome aneuploidies as compared to Karyotyping.
- QF-PCR detection of aneuploidies is a well established technique. The data presented here confirms that the Devyser® Complete kit is reliable, accurate and robust with no misdiagnoses for non-mosaic trisomy 13, 18 or 21, Klinefelter, Turner or triploidy in this material.
- The Devyser® Complete QF-PCR kit allows true detection of Turner syndrome.

Figure 1. Typical electrophoretogram (Mix 1) showing the profile of a trisomic sample (47, XY +21)



Introduction

Devyser® Complete is a CE-labelled IVD kit for prenatal diagnosis of the most common autosomal and sex chromosome aneuploidies, including Down, Edward, Patau, Klinefelter and Turner syndromes. A high number of markers, seven markers each for chromosomes 13, 18, 21 and X/Y, are included to significantly reduce the need for re-runs. The kit includes ready-to-use reagents and is delivered with the Devyser® Decision Base software.



Materials and Methods

A total of 333 amniotic fluid (AF) samples and 184 whole blood samples were analysed in order to evaluate the clinical usefulness, marker informativity and correlation to karyotyping.

QIAamp DNA Blood Kit (Qiagen) and InstaGene Matrix (Bio-Rad) were used for manual DNA extraction of AF samples. MagNA Pure LC DNA Isolation Kit I was used for automated DNA extraction from whole blood samples. The Devyser® Complete QF-PCR kit was used according to the Instructions for Use. PCR amplification was performed using ABI GeneAmp® System 9700, Eppendorf Mastercycler or MJ Research PTC200. Detection was performed using ABI PRISM® 3700, ABI PRISM® 3100 or ABI PRISM® 3130.

Results

Out of the 333 amniotic fluid samples analyzed, 319 were normal. Totally 14 samples showed autosomal or sex chromosome aneuploidies (see table 1 for details). Three samples exhibited maternal cell contamination and could not be analyzed using QF-PCR. In total, 330 samples gave analyzable results using Devyser® Complete showing 100% specificity and sensitivity with results previously obtained by karyotyping. Results from the Devyser® Complete QF-PCR analysis of 333 amniotic fluid samples are summarized in table 1. Atypical Electrophoretogram is shown in figure 1.

Table 1. Results from testing of 333 amniotic fluid samples.

Diagnosis	Number of Samples	
	Devyser® Complete	Karyotyping
Normal	316	319
Trisomy 13 (Patau)	1	1
Trisomy 18 (Edward)	1	1
Trisomy 21 (Down)	7	7
Triploid	1	1
Turner (X0)	2	2
Klinefelter (XXY)	2	2
Significant maternal cell contamination*	3	NA
Failure to amplify	0	NA
Uninformative	0	0
Total	333	333

NA = Not Applicable

*) Maternal blood sample was not available and sample could not be resolved

Marker informativity rates were calculated from a total of 515 samples (333 amniotic fluid samples and 182 whole blood samples) and ranged from 67 - 100 % in the tested samples

