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RAPORT ANALIZE MEDICALE

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Raport de analize medicale nr. 2094

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Nume: **ENCULESCU EMA**	Varsta: **0** Sex: **F**	Medic: STAN IUSTINA
C.N.P:	Cod pacient: **6677**	CONTRACT: **PLATA - iunie/2010**

GENETICA MOLECULARA *- LUCRAT DE : CRISTINA DRAGOMIR - BIOCHIMIST SPECIALIST*

Detectia mutatiilor genei CFTR - Fibroza chistica (32 mutatii)

Testul are capacitatea de a detecta urmatoarele mutatii in forma heterozigota sau homozigota, in gena CFTR: R347P, 2789+5G>A, 3120+1G>A, 711+1G>T, R334W, I507del, F508del, 3849+10kbC>T, 1078delT, W1282X, R560T, R553X, G551D, S549RT>G, M1101K, G542X, 3905insT, S1251N, 1717-1G>A, R117H, N1303K, G85E, 1898+1G>A, 2184delA, D1152H, CFTRdele2,3, P67L, E60X, 3659delC, 621+1G>T, A455E, R1162X

REZULTAT: S-a detectat mutatia 2184delA in forma heterozigota.