UNUSUAL FINDING IN A PATERNITY TESTING CASE WITH AN XX ALLEGED FATHER

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During a routine paternity testing case, a paternal exclusion was found. However, it called our attention that the alleged father was an XX individual with Amelogenin.

Locus	AF	Child	Mother
FGA	19 / 20	20 / 24	24 / 24
TPOX	11 / 12	8 / 9	11 / 8
D8S1179	13 / 15	13 / 14	13 / 14
VWA	16 / 18	16 / 18	16 / 17
D18S51	13 / 16	13 / 13	13 / 13
D21S11	30 / 30	30 / 31	31 / 32.2
TH01	6 / 9	9 / 9	6 / 9
D3S1358	16 / 17	15 / 17	15 / 15
CSF1PO	10 / 9	11 / 12	10 / 12
D16S539	11 / 12	11 / 14	11 / 14
D7S820	12 / 13	8 / 9	10 / 9
D13S317	11 / 12	12 / 8	11 / 8
D5S818	11 / 12	12 / 12	11 / 12
D19S433	12 / 14	13 / 13	12 / 13
D2S1338	21 / 23	20 / 24	20 / 20
Amelogenina	XX	XX	XX

A new sample from the alleged father was requested. Once we received the new sample, we were informed that the alleged father had received a Bone Marrow transplantation (BMT) eight years before. His HLA identical sister was the donor for the BMT. We contacted the alleged father in order to obtain a mouth cell sample. Once the sample was analyzed, a mixture XY profile was obtained. In the mixture, the obligated paternal alleles that did not exclude the paternity were found. We thought that the sample was a traumatic sample and we proceeded to take new samples avoiding any trauma. Three different samples were obtained, one from the inner right cheek, one from the inner left cheek and one from the tongue.

Two of the three samples showed an identical genetic XX profile as found in the first blood sample without evidence of the XY profile. In the third sample, an XY genetic profile was found with minor peaks for some alleles found in the XX profile.

Locus	A.F (B)	AF. MC # 1	AF new MC#1	AF new MC#2	AF new MC#3
FGA	19 / 20	19 / 20	19 / 20	19 / 20	19 / 20
TPOX	11 / 12	(9)/ (11) / 12	11 / 12	9 / 12	11 / 12
D8S1179	13 / 15	8 / 13 / (15)	13 / 15	8 / 13	13 / 15
VWA	16 / 18	16 / 18	16 / 18	16 / 18	16 / 18
D18S51	13 / 16	13 / (14)/ (16)	13 / 16	13 / 14	13 / 16
D21S11	30 / 30	30 / 30	30 / 30	30 / 30	30 / 30
TH01	6 / 9	6 / 9	6 / 9	6 / 9	6 / 9
D3S1358	16 / 17	16 / 17	16 / 17	16 / 17	16 / 17
CSF1PO	10 / 9	11 / (10) / (9)	10 / 9	11/ 11	10 / 9

D16S539	11 / 12	9 / 11 / (12)	11 / 12	9 / 11	11 / 12
D7S820	12 / 13	8 / 11 / 12 / 13	12 / 13	11 / 8	12 / 13
D13S317	11 / 12	11 / 12	11 / 12	11 / 12	11 / 12
D5S818	11 / 12	11 / 12	11 / 12	11 / 12	11 / 12
D19S433	12 / 14	(12) / 13 / (14)	12 / 14	13 / 13	12 / 14
D2S1338	21 / 23	21 / (23) / (24)	21 / 23	21 / 24	21 / 23
Amelogenina	XX	X(Y)	XX	XY	XX

AF: Alleged Father. B: Blood Sample. MC: Mouth cells.

This case had been previously studied in a different laboratory and was reported as an exclusion of paternity. No mention to the XX male profile was reported.

Our results indicate that the post-transplant chimerism was not limited to the bone marrow derived cells. The stem cells from the HLA identical donor had also replaced a good part of the mouth cells mucosa. No other samples were available for testing.

Recently, after a liver transplantation case in Australia, the recipient's bone marrow was replaced with donor specific stem cells. In addition, several reports in the literature had described genetic microchimerisms after kidney transplantation and blood transfusions.

Our results indicate that in paternity testing cases where one of the parties had received a bone marrow transplantation, exhaustive studies should be carried out, even more, if genetic inconsistencies are found as those described in the present study.