

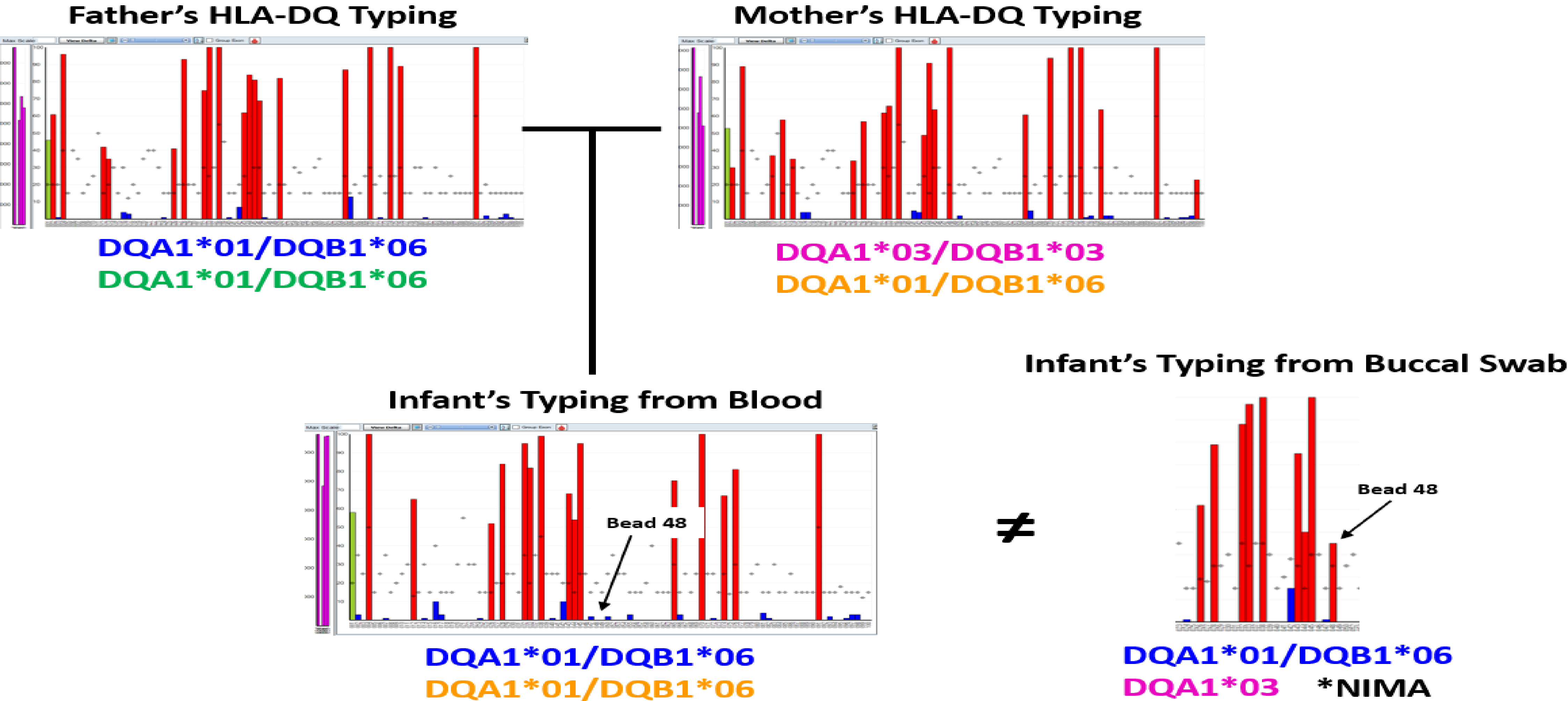
Introduction

- A one-month-old infant was evaluated as a potential donor for their sibling.
- A buccal swab sample was collected two hours after breast feeding to limit maternal DNA contamination.
- HLA typing was performed by Next Generation Sequencing.
- The infant’s HLA-DQA1 (DQA1*01) and DQB1 (DQB1*06) typings were homozygous and re-tested using a second method.
- The rSSO HLA-DQA1 results were discordant (DQA1*01, DQA1*03) showing heterozygosity at the locus.

Methods

- Bead 48, which is specific for DQA1*03, was near the manufacturer’s cut-off and upon rSSO repeat, was under the cut-off suggesting low level contamination of the buccal swab sample.
- The HLA-DQA1 and DQB1 typings of the father and mother were performed, and the heterozygous result was confirmed as breast milk contamination by the non-inherited HLA-DQA1 maternal allele.
- Subsequently, a blood sample was used to verify the infant’s HLA-DQA1 homozygosity.

Results



Conclusions

- An attempt to reproduce these results was performed with a lab technologist and her infant. Infant buccal swabs were purposely collected at various time intervals after breast feeding and rSSO performed on both the mother and infant. No maternal antigens were detected. However, she is a match with her infant son at DQA1*01, leading us to conclude that the primer combination may have been involved in the detection of the non-inherited maternal HLA allele.
- It has been previously recommended to collect infant buccal swab samples one hour post breast feeding to avoid contamination by non-inherited maternal antigens.¹
- However, buccal swabs collected two hours after breast feeding still resulted in a discordant HLA typing. Even though NGS filtered out the contamination and rSSO picked it up marginally, the potential for mistyping is clear.
- While a buccal swab is a non-invasive method of sample collection for infants, inquiry of an infant’s breast feeding status is important to obtain accurate laboratory results. We recommend prior to sample collection to wipe the mucosal surface with cotton applicator.
- 1. Babovic-Vuksanovic D, Michels VV, Law ME, Bailey R, Wyatt WA, Lindor NM, Jalal SM: Guidelines for buccal smear collection in breast-fed infants. Am J Med Genet 84:4, 1999.