OBJECTIVE:
To add evidence that massive parallel sequencing (MPS) is a valuable substitute for array comparative genomic hybridization with a resolution that is more appropriate for preimplantation genetic diagnosis in translocation carriers.

DESIGN:
Study of diagnostic accuracy.

SETTING:
University hospital setting.

PATIENT(S):
Fifteen patients with a balanced structural rearrangement were included in the study: eight reciprocal translocations, four Robertsonian translocations, two inversions, and one insertional translocation.

INTERVENTION(S):
Trophectoderm biopsy was performed on 47 blastocysts.

MAIN OUTCOME MEASURE(S):
In the current study, shallow whole genome MPS on a NextSeq500 (Illumina) and Ion Proton (Life Technologies) instrument was performed in parallel on 47 WGA amplified trophoderm samples. Data analyses were performed using the QDNAseq algorithm implemented in Vivar.

RESULT(S):
In total, 5 normal and 42 abnormal embryos were analyzed. All aberrations previously detected with array comparative genomic hybridization could be readily detected in the MPS data using both technologies and were correctly identified. The smallest detected abnormality was a ∼4.5 Mb deletion/duplication.

CONCLUSION(S):
This study demonstrates that shallow whole genome sequencing can be applied efficiently for the detection of numerical and structural chromosomal aberrations in embryos, equaling or even exceeding the resolution of the routinely used microarrays.