

## **Abstract**

The normal range of activities of 6 lysosomal enzymes was determined in extracts of chorionic villi samples obtained by a rigid forceps in the first trimester of pregnancy. These activities were compared to those in villi obtained after abortion and in cultured amniotic fluid cells and fibroblasts. For five of the six enzymes tested, the data suggest that first trimester prenatal diagnosis should be possible and reliable. For the sixth enzyme, alpha-L-fucosidase, where on occasion very low activities were found, the results obtained on fresh chorionic villi have to be interpreted with extreme caution. Considerable lysosomal enzyme activities were also found in maternal decidua. Therefore, extreme care must be taken in the preparation of chorionic villi for prenatal diagnosis of lysosomal disorders since even small amounts of maternal tissue could lead to misdiagnosis. This study has allowed us to monitor 2 pregnancies at risk for lysosomal storage disease. Differences in the isoenzyme pattern of alpha-L-fucosidase were found in chorionic villi and maternal decidua. Although further studies are required, this observation could lead to the development of immuno-biochemical methods to evaluate the purity of chorionic villi used for prenatal diagnosis.