

SP prodomain ASD

Ala249Glu Ala319Thr

c.746C>A; p.Ala249Glu

patient 792-535

Ala Arg Ala>Glu Arg Gly
G C G C G C G C G A C G G G G A

c.995G>A; p.Ala319Thr

patient ID178

Ser Gly Ala>Thr Pro Asp
T C G G G C G C C C C G G A T

Ala249Glu/+ +/+

792-535
Ala249Glu/+ +/+