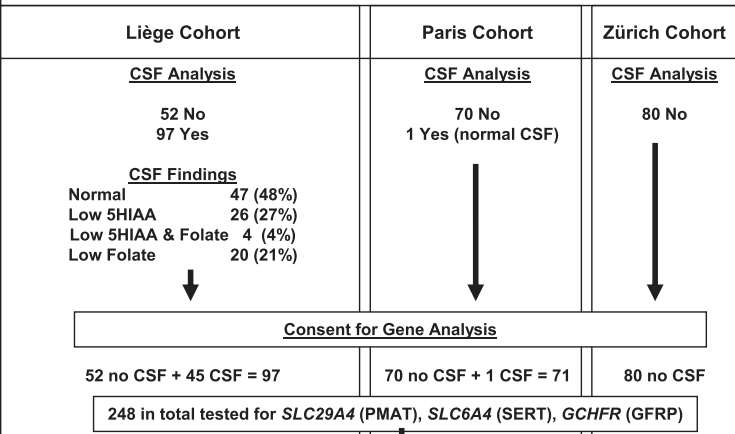


Diagnostic Flow of ASD in 248 Caucasian Patients



Genetic Findings heterozygous; 248 ASD subjects from all three cohorts (= 100%)

- 5 with SNP rs73332823/p.M24L (PMAT) (2%)
- 2 with c.86A>G/p.D29G (PMAT) (0.8%)
- 5 with c.412G>A/p.A138T (PMAT) (2%)
- 1 with c.978T>G/p.D326E (PMAT)* (0.4%)
- 5 with SNP rs6355/p.G56A (SERT)* (2%)

Mutant PMAT expression and functional studies in MDCK cells

Controls

