### Diagnostic Flow of ASD in 248 Caucasian Patients

<table>
<thead>
<tr>
<th>Liège Cohort</th>
<th>Paris Cohort</th>
<th>Zürich Cohort</th>
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<tbody>
<tr>
<td><strong>CSF Analysis</strong></td>
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<tr>
<td>52 No 97 Yes</td>
<td>70 No 1 Yes (normal CSF)</td>
<td>80 No</td>
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<tr>
<td><strong>CSF Findings</strong></td>
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<td>Normal 47 (48%)</td>
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<tr>
<td>Low 5HIAA 26 (27%)</td>
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<tr>
<td>Low 5HIAA &amp; Folate 4 (4%)</td>
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<td>Low Folate 20 (21%)</td>
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Consent for Gene Analysis

- 52 no CSF + 45 CSF = 97
- 70 no CSF + 1 CSF = 71
- 80 no CSF

248 in total tested for SLC29A4 (PMAT), SLC6A4 (SERT), GCIR (GFRP)

### 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

- (no CSF Analysis)

- Gene Analysis in total 15,982 Chromosomes (= 100%)
  - 394 Unaffected Caucasian Controls (788 Chromosomes**)
    - 1,000 Genome Project (2,188 Chromosomes***)
    - Exome Sequence Project (13,006 Chromosomes****)

- Genetic Findings (heterozygous)
  - SNP rs73332823/p.M24L (PMAT) (1.9%)
    - 23 with c.86A>G/p.D29G (PMAT) (0.14%)
    - 0 with c.412G>A/p.A138T (PMAT) (<0.007%)
    - 0 with c.978T>G/p.D326E (PMAT) (<0.007%)

- SNP rs6355/p.G56A (SERT) (2.3%) ref. 25