

| Mutations status             | Subject | Gender | KS* | Neonatal RD | Recurrent upper respiratory tract infections | Bronchiectasis Broncho-pneumonia | Nasal polyps | Reduced mucociliary clearance | Absence Ciliary Movement |
|------------------------------|---------|--------|-----|-------------|--|----------------------------------|--------------|-------------------------------|--------------------------|
| <b>Homozygous</b>            | 8227    | F      | Yes | No          | Yes  | Yes                              | N/A          | N/A                           | N/A                      |
| <b>Compound Heterozygous</b> | 6181    | F      | No  | Yes         | Yes  | Yes                              | No           | N/A                           | Yes                      |
| <b>Heterozygous</b>          | 6127    | M      | No  | Yes         | Yes  | Yes                              | No           | No                            | Yes                      |
|                              | 6177    | M      | No  | No          | Yes  | Yes                              | Yes          | Yes                           | Yes                      |

**Table E2: Clinical details of the 4 patients carrying *DNAI1* mutations**

There was no known parental consanguinity in any of these families.

\*When a *situs inversus* was observed, patients were referred as having Kartagener Syndrome.

Abbreviations: M: male, F: female, KS: Kartagener Syndrome, RD: respiratory distress, N/A: not available.