Supplemental Table 1. Profiles of 131 patients at initial CMA testing

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| --- | --- | --- | --- |
| Characteristics | | N=131 | (%) |
| Sex | Male | 65 | 49.6 |
|  | Female | 66 | 50.4 |
| Agea | < 1 month b | 2 | 1.5 |
|  | 1-12 month | 46 | 35.1 |
|  | 1-6 years | 57 | 43.5 |
|  | 7-12 years | 18 | 13.7 |
|  | > 13 years c | 8 | 6.1 |
| Indication for CMA testing d | DD/ID | 15 | 11.5 |
|  | MCA/DD | 113 | 86.3 |
|  | epilepsy | 3 | 2.3 |
| G-banding | normal | 103 | 78.6 |
|  | abnormal | 27 | 20.6 |
|  | unexamined | 1 | 0.8 |
| Array platforme for CMA | CytoScan HD | 113 | 86.3 |
|  | CytoScan 750K | 18 | 13.7 |
| Interpretation d | Pathogenic/Likely Pathogenic | 22 | 16.8 |
|  | Unsolved | 109 | 83.2 |

DD: development disability, ID: intellectual disability, MCA: multiple Congenital Disorder

a: mean of three years and six months, a median of one year and five months

b: two days is the minimum age included in the <1 age classification

c: 19 years old is the maximum age included in the >13 age classification

d: at the initial analysis by 400 kb and 50 probes threshold