**Supplementary table 1a.** Characteristics of all individual subjects

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Subject | Sex | Mutation (gene) | cDNA level | Protein level | CA  baseline | CA  1 year | CA 2 year | CA after suspension | BA  baseline | BA  1 year | BA 2 year |
| 1 | F | KMT2D | c.5912delG | p.Ser1971Thrfs\*76 | 9,42 | 10,52 | 11,78 | - | 8,00 | 9,50 | 11,00 |
| 2 | M | KMT2D | c.8200C>T | p.Arg2734\* | 8,82 | 9,92 | 11,01 | 12,38 | 9,00 | 10,00 | 11,50 |
| 3 | F | KMT2D | c.9770dupA | p.Lys3258Glufs\*43 | 6,80 | 7,84 | 8,84 | 9,81 | 5,75 | 6,83 | 7,83 |
| 4 | F | KMT2D | c.12969dupA | p.Pro4324Thrfs\*10 | 5,94 | 6,99 | 8,11 | 9,01 | 4,00 | 4,50 | 6,00 |
| 5 | F | KMT2D | c.11497delC | pGlyfsx48 | 5,92 | 7,06 | 8,07 | 8,91 | 5,75 | 6,83 | 7,83 |
| 6 | M | KMT2D | c.4209C>G | p.Cys1403Trp | 7,09 | 8,23 | 9,23 | 10,11 | 7,00 | 8,50 | - |
| 7 | F | KMT2D | c.11566C>T | p.Gln3856\* | 8,07 | 9,20 | 10,08 | 11,19 | 6,83 | 7,83 | 9,00 |
| 8 | M | KMT2D | c.2129delC | p.Pro710Hisfs\*220 | 4,02 | 5,04 | 6,00 | 6,99 | 3,00 | 4,00 | 6,00 |
| 9 | M | KMT2D | c.10834C>T | p.Glu3612x | 10,07 | 11,18 | 12,25 | 12,42 | 9,00 | 12,50 | 13,00 |
| 10 | M | KMT2D | c.3307\_3310del | p.Cys1103Profs\*15 | 4,17 | 5,18 | 6,18 | 7,83 | 3,00 | 4,50 | 6,00 |
| 11 | M | KMT2D | c.15142C>T | p.Arg5048Cys | 6,01 | 7,01 | 8,05 | 9,34 | 4,00 | 5,50 | 6,00 |
| 12 | F | KMT2D | c.16294C>T | p.Arg5432Trp | 4,43 | 5,43 | 6,52 | 7,44 | 3,00 | 5,00 | 6,83 |
| 13 | F | KMT2D | c.16018C>T | p.Arg5340x | 5,12 | 6,10 | 7,10 | 8,36 | 5,00 | 6,50 | 6,83 |
| 14 | M | KDM6A | KMD6A deletie exon 6 | - | 9,89 | 10,90 | 11,82 | 12,58 | 8,00 | 9,00 | 10,00 |
| 15 | F | KDM6A | KMD6A VUS exon 11 | - | 3,78 | 4,89 | 5,76 | 6,67 | 3,00 | 4,17 | 5,00 |
| 16 | M | KMT2D | c.4871C>T | r.4870\_4963del67; p.Ala1624Val | 7,53 | 8,52 | 9,65 | - | 7,00 | 8,00 | 10,00 |
| 17 | M | KMT2D | c.4207T>C | p.Cys1403Arg | 9,47 | 10,60 | 11,62 | - | 7,00 | 9,00 | 11,00 |
| 18 | F | KDM6A | c.2243C>T | p.Pro748Leu | 7,82 | 8,90 | 9,89 | 10,73 | 7,83 | 8,83 | 11,00 |

CA, calendar age in years; BA, bone age in years

**Supplementary table 1b.** Characteristics of all individual subjects

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Subject | HSDS  baseline | HSDS  1 year | HSDS  2 year | HSDS  After suspension | BMISDS  baseline | BMISDS  1 year | BMISDS  2 year | BMISDS  After suspension | GHD (peak range clonidine/arginine µg/l)\* |
| 1\* | -1,15 | -1,08 | -,87 | - | 2,76 | 2,08 | 2,10 | - | - (8.33/6.03) |
| 2 | -5,36 | -4,98 | -4,38 | -4,42 | ,54 | -,52 | -,52 | ,46 | - (1.37/13.33) |
| 3\* | -,56 | ,29 | ,90 | ,24 | 2,93 | 2,61 | 2,29 | 2,98 | + (4.30/3.80) |
| 4 | -1,83 | -1,07 | ,07 | -,42 | -,36 | -,17 | -,50 | -,09 | - (12.07/12.07) |
| 5 | -1,29 | -,39 | ,28 | ,02 | ,05 | -,32 | -1,30 | -,26 | + (6.73/5.27) |
| 6 | -1,02 | -,29 | -,04 | -,15 | -,60 | -1,06 | -,84 | - | - (9.70/8.30) |
| 7 | -5,81 | -5,24 | -4,81 | -4,48 | ,06 | ,30 | ,38 | ,65 | - (5.47/11.80) |
| 8 | -2,78 | -1,25 | -,96 | ,01 | 1,63 | ,35 | 1,00 | ,09 | + (5.93/5.20) |
| 9 | -1,52 | -,80 | -,63 | -,78 | ,09 | ,73 | ,40 | ,92 | - (31.93/x) |
| 10 | -3,82 | -2,77 | -2,30 | -2,63 | 2,05 | ,78 | ,58 | 2,20 | - (15.03/8.80) |
| 11 | -2,59 | -1,77 | -1,42 | -1,86 | -2,32 | -1,57 | -1,97 | -,84 | - (17.83/x) |
| 12\* | -2,60 | -1,82 | -1,75 | -1,86 | -1,91 | -1,68 | -1,46 | -,75 | + (5.30/6.27) |
| 13 | -1,22 | -,15 | ,32 | ,00 | -,31 | ,79 | ,80 | 1,34 | - (20.07/6.23) |
| 14\* | -6,19 | -5,78 | -5,20 | -5,27 | -1,48 | -2,21 | -2,05 | -3,22 | No data, done elsewhere |
| 15 | -2,48 | -2,02 | -1,52 | -1,76 | -,72 | -1,22 | -,08 | ,17 | - (x/15.47) |
| 16\* | -1,09 | -,63 | -,35 | - | 4,31 | 2,42 | 1,98 | - | - (4.60/7.33) |
| 17 | ,85 | 1,37 | 1,21 | - | 1,58 | ,97 | ,91 | - | - (5.63/11.60) |
| 18\* | -2,72 | -2,08 | -1,90 | -2,11 | 1,80 | 1,06 | 1,51 | 1,69 | - (7.03/8.43) |

\*, subjects with IGF-I SDS levels >2 during treatment

H, height in SDS; GHD, growth hormone deficiency