

Additional file 3.

Title: Karyotypic groups for statistical characterization of the cohort

Legend: Groups were formed according to the nature of chromosome abnormality (numerical/structural, pure/mosaic, loss/gain of chromosomal material, type of sex chromosome aberration/gonadal dysgenesis). Parentheses involve the total number of patients in the group, slashes the concrete patients. Colon indicates an interval.

SCA and OGD groups for statistical characterization (with exception of birth weight and length) of the cohort:

1. All SCAs and OGDs (137) /P1:137/
2. Non-mosaic SCAs and OGDs (102) /P1:19;40;42:44;47:51;53:75;78;81:87;89:100;102:132/
3. Mosaic SCAs and OGDs (35) /P20:39;41;45:46;52;76:77;79:80;88;101;133:137/
4. Non-structural SCAs and OGDs (90) /P1:39;53:79;82:88;90:101;133:137/
5. Structural SCAs and OGDs (47) /P40:52;80:81;89;102:132/
6. SCAs and OGDs without quantitative alterations (20) /P104:106;117:133/
7. SCAs and OGDs with quantitative alterations (117) /P1:103;107:116;134:137/
8. SCAs and OGDs with loss of SC material (47) /P1:36;40:45;103;134:137/
9. SCAs and OGDs with gain of SC material (50) /P53:101;107/
10. SCAs and OGDs with combined quantitative alterations (20) /P37:39;46:52;102;108:116/
11. Non-mosaic, non-structural SCAs and OGDs (60) /P1:19;53:75;78;82:87;90:100/
12. Non-mosaic, structural SCAs and OGDs (42) /P40;42:44;47:51;81;89;102:132/
13. Mosaic, non-structural SCAs and OGDs (30) /P20:39;76:77;79;88;101;133:137/
14. Mosaic, structural SCAs and OGDs (5) /P41;45:46;52;80/
15. Non-mosaic, non-structural loss of SC material (19) /P1:19/
16. Non-mosaic, non-structural gain of SC material (41) /P53:75;78;82:87;90:100/
17. Non-mosaic, structural SCAs and OGDs without quantitative alterations (19)
/P104:106;117:132/
18. Non-mosaic, structural SCAs and OGDs with quantitative alterations (23)
/P40;42:44;47:51;81;89;102:103;107:116/
19. Non-mosaic, structural loss of SC material (5) /P40;42:44;103/
20. Non-mosaic, structural gain of SC material (3) /P81;89;107/

21. Non-mosaic, structural SCAs and OGDs with combined quantitative alterations (15)
/P47:51;102;108:116/
22. Mosaic, non-structural SCAs and OGDs without quantitative alterations (1) /P133/
23. Mosaic, non-structural SCAs and OGDs with quantitative alterations (29)
/P20:39;76:77;79;88;101;134:137/
24. Mosaic, non-structural loss of SC material (21) /P20:36;134:137/
25. Mosaic, non-structural gain of SC material (5) /P76:77;79;88;101/
26. Mosaic, non-structural SCAs and OGDs with combined quantitative alterations (3)
/P37:39/
27. Mosaic, structural SCAs and OGDs without quantitative alterations (0) /P-/
28. Mosaic, structural SCAs and OGDs with quantitative alterations (5) /P41;45:46;52;80/
29. Mosaic, structural loss of SC material (2) /P41;45/
30. Mosaic, structural gain of SC material (1) /P80/
31. Mosaic, structural SCAs and OGDs with combined quantitative alterations (2) /P46;52/
32. Turner sy. and variants (52) /P1:52/
33. Klinefelter sy. and variants (29) /P53:81/
34. Triple/Poly X sy. and variants (9) /P82:90/
35. Double Y sy. (11) /P91:101/
36. Combined partial deletion and duplication of X chromosome (1) /P102/
37. Partial Y deletion (1) /P103/
38. X-autosome translocations (4) /P104:107/
39. X-Y translocations (9) /P108:116/
40. Other gonadal dysgeneses (21) /P117:137/

SCA and OGD groups for statistical characterization of the cohort's birth weight and length:

1. All SCAs and OGDs (103)
/P1;3:4;6:14;16:34;36:38;41:42;44:49;51:56;62:64;66;70:71;73:75;77:85;88:96;98;104:108;10:115;117;120:121;123:130;133:137/
2. Non-mosaic SCAs and OGDs (72)
/P1;3:4;6:14;16:19;42;44;47:49;51;53:56;62:64;66;70:71;73:75;78;81:85;89:96;98;104:108;10:115;117;120:121;123:130/

3. Mosaic SCAs and OGDs (31) /P20:34;36:38;41;45:46;52;77;79:80;88;133:137/
4. Non-structural SCAs and OGDs (68)
/P1;3:4;6:14;16:34;36:38;53:56;62:64;66;70:71;73:75;77:79;82:85;88;90:96;98;133:137/
5. Structural SCAs and OGDs (35)
/P41:42;44:49;51:52;80:81;89;104:108;110:115;117;120:121;123:130/
6. SCAs and OGDs without quantitative alterations (15)
/P104:106;117;120:121;123:130;133/
7. SCAs and OGDs with quantitative alterations (88)
/P1;3:4;6:14;16:34;36:38;41:42;44:49;51:56;62:64;66;70:71;73:75;77:85;88:96;98;107:108;110:115;134:137/
8. SCAs and OGDs with loss of SC material (40) /P1;3:4;6:14;16:34;36;41:42;44:45;134:137/
9. SCAs and OGDs with gain of SC material (33)
/P53:56;62:64;66;70:71;73:75;77:85;88:96;98;107/
10. SCAs and OGDs with combined quantitative alterations (15)
/P37:38;46:49;51:52;108;110:115/
11. Non-mosaic, non-structural SCAs and OGDs (42)
/P1;3:4;6:14;16:19;53:56;62:64;66;70:71;73:75;78;82:85;90:96;98/
12. Non-mosaic, structural SCAs and OGDs (30)
/P42;44;47:49;51;81;89;104:108;110:115;117;120:121;123:130/
13. Mosaic, non-structural SCAs and OGDs (26) /P20:34;36:38;77;79;88;133:137/
14. Mosaic, structural SCAs and OGDs (5) /P41;45:46;52;80/
15. Non-mosaic, non-structural loss of SC material (16) /P1;3:4;6:14;16:19/
16. Non-mosaic, non-structural gain of SC material (26)
/P53:56;62:64;66;70:71;73:75;78;82:85;90:96;98/
17. Non-mosaic, structural SCAs and OGDs without quantitative alterations (14)
/P104:106;117;120:121;123:130/
18. Non-mosaic, structural SCAs and OGDs with quantitative alterations (16)
/P42;44;47:49;51;81;89;107:108;110:115/
19. Non-mosaic, structural loss of SC material (2) /P42;44/
20. Non-mosaic, structural gain of SC material (3) /P81;89;107/
21. Non-mosaic, structural SCAs and OGDs with combined quantitative alterations (11)
/P47:49;51;108;110:115/

22. Mosaic, non-structural SCAs and OGDs without quantitative alterations (1) /P133/
23. Mosaic, non-structural SCAs and OGDs with quantitative alterations (25)
/P20:34;36:38;77;79;88;134:137/
24. Mosaic, non-structural loss of SC material (20) /P20:34;36;134:137/
25. Mosaic, non-structural gain of SC material (3) /P77;79;88/
26. Mosaic, non-structural SCAs and OGDs with combined quantitative alterations (2)
/P37:38/
27. Mosaic, structural SCAs and OGDs without quantitative alterations (0) /P-/
28. Mosaic, structural SCAs and OGDs with quantitative alterations (5) /P41;45:46;52;80/
29. Mosaic, structural loss of SC material (2) /P41;45/
30. Mosaic, structural gain of SC material (1) /P80/
31. Mosaic, structural SCAs and OGDs with combined quantitative alterations (2) /P46;52/
32. Turner sy. and variants (44) /P1;3:4;6:14;16:34;36:38;41:42;44:49;51:52/
33. Klinefelter sy. and variants (18) /P53:56;62:64;66;70:71;73:75;77:81/
34. Triple/Poly X sy. and variants (7) /P82:85;88:90/
35. Double Y sy. (7) /P91:96;98/
36. Combined partial deletion and duplication of X chromosome (0) /P-/
37. Partial Y deletion (0) /P-/
38. X-autosome translocations (4) /P104:107/
39. X-Y translocations (7) /P108;110:115/
40. Other gonadal dysgeneses (16) /P117;120:121;123:130;133:137/

Abbreviations: OGD: other gonadal dysgenesis; p: short arm of a chromosome; P: patient; q: long arm of a chromosome; SC: sex chromosomal; SCA: sex chromosome abnormality; sy.: syndrome; v.: variant; X: X chromosome; Y: Y chromosome