

Supplementary Figure 1: Cell population-based SUMOylation pattern distribution in the individual phenotypes.

Pachytene cells analyzed from four individuals were categorized based on the observed small ubiquitin-related modifier (SUMO)-1 (panel I) or SUMO-2/3 (panel II) pattern. Each chart is based on the findings for 100% of pachytene cells analyzed from each patient (A-D).

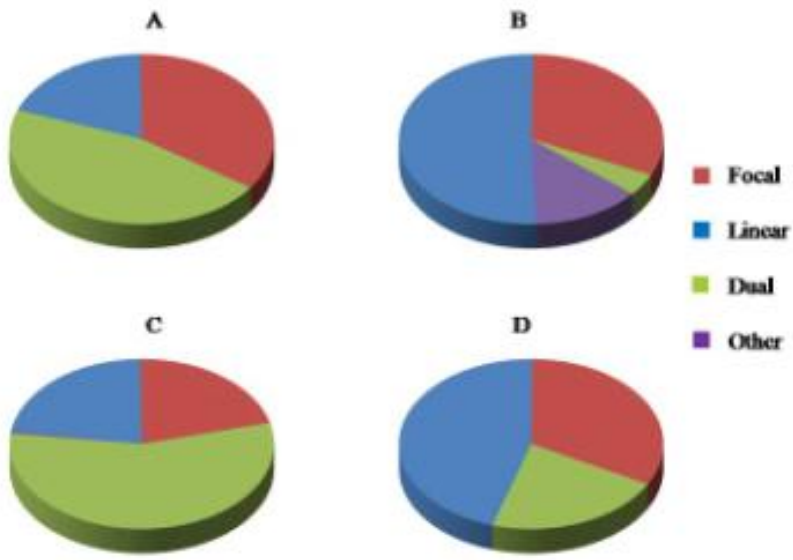
A: Control. Normal; obstructed (congenital bilateral absence of the vas deferens), absence of cystic fibrosis disease

B: hypospermatogenesis

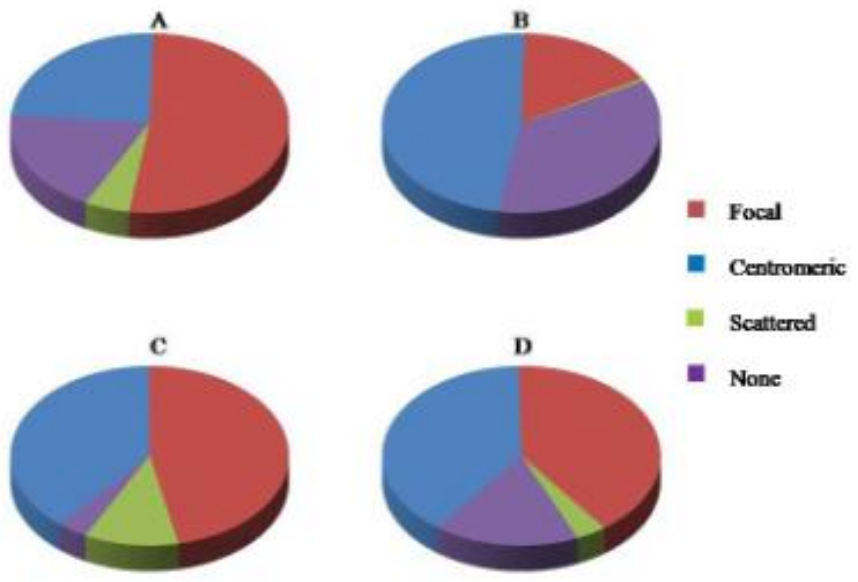
C: hypogonadotropic hypogonadism (Kallmann syndrome)

D: Normal spermatogenesis in tissue from patient with a seminoma

I. SUMO-1



II. SUMO-2/3



Statistical analysis

To test for statistical significance in the altered frequencies in the patterns observed within our sample population for both SUMO-1 and SUMO-2/3, separately, we used the Chi-square test. There is a significant difference between the incidence of the three SUMO-1 patterns and “other” between the four individuals ($\chi^2 = 164.56$; $p < 0.0001$). When comparing the three infertile cases with normal spermatogenesis (Patient A), Patient B ($\chi^2 = 72.0$; $p < 0.0001$) and Patient D were highly significantly different ($\chi^2 = 25.5$; $p = 2.9 \times 10^{-6}$). Patient C was not significantly different from Patient A ($\chi^2 = 7.1$; $p = 0.069$). When comparing the four SUMO-2/3 patterns, including “none”, a significant difference was observed between the four individuals ($\chi^2 = 136.31$; $p < 0.0001$). When comparing the three cases with the control, Patients B, C, and D were significantly different ($\chi^2 = 76.48$; $p < 0.0001$; $\chi^2 = 10.28$; $p = 0.016$; $\chi^2 = 34.10$; $p = 1.9 \times 10^{-7}$, respectively).