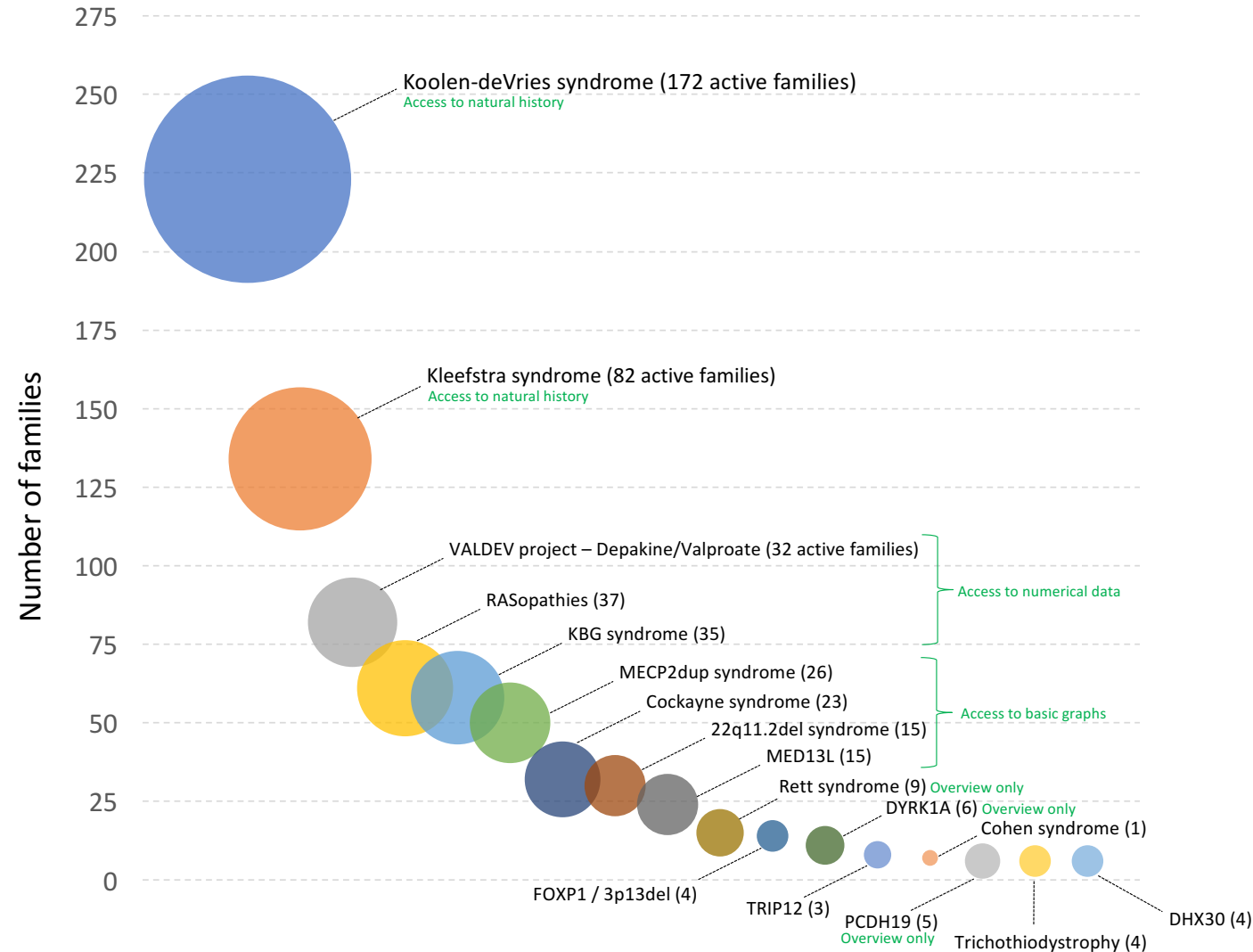


## Patients groups in GENIDA – Sept. 2018



## Smaller cohorts in GENIDA

2 to 6 patients

15q11.2 duplication - 16p11.2 deletion syndrome - 17p13.3 triplication cohort - 18p tetrasomy syndrome - 1p36 deletion cohort - 48,XXXY syndrome - 9q33.3q34.11 deletion cohort - ADNP syndrome - CDKL5 cohort - CHAMP1 cohort - CNKSR2 cohort - Cri du chat syndrome - DEAF1 cohort - Down Syndrome / T21 - FOXG1 cohort - GATAD2B cohort - Glass syndrome - GRIN2B cohort - HCN1 cohort - KCNQ2 cohort - KCNT1 cohort - KIAA2022 cohort - KMT2C cohort - MYT1L cohort - Phelan-McDermid syndrome - PHIP cohort - Pitt-Hopkins syndrome - SETBP1 cohort - SETD5 cohort - STXBP1 cohort - SYNGAP1 cohort - TBCK cohort - TRIO cohort - Wiedemann-Steiner syndrome - ZSWIM6 cohort

## Other genetic forms in GENIDA

ACSL4 - AHDC1 - ARX - ASXL3 - BCL11A - BRPF1 - CCND2 - CDK13 - CNKSR2 - DEAF1 - FMR1 - GABRG2 - KMT2C - KMT2D - MLS3 - MYT1L - NACC1 - NOTCH1 - NR2F1 - PHIP - PRRT2 - RAI1 - RNF113A - RPS6KA3 - SHANK2 - SHOC2 - SLC6A8 - SNC8A - SOX5 - STAG1 - STXBP1 - TRAPPC9 - UBE3A - USP7 - ZSWIM6

1p35.1del - 1p36.3 to 3q29 unbalanced transl. - 1q21.1dup - 1q21.2q21.3del - 2p16.3del - 2q23.3dup - 2q24.1-24.2dup - 2q26.3del - 2q31.1del - 2q32.3q33.1del - 2q37del - 2q37.3del - 2q37.3dup - 3p13p12.3dup - 3q13.31del - 3q28q29dup - 3qterdel - 4pdup - 4p16.3dup - 4q21del - 5p12 to 1q31 unbalanced transl. - 5p15.32p15.33del - 5q14.3del - 6p24.1p22.3del - 6p25.3dup - 6q14.1del - 6q26dup - 7p21.2del - 7q31.33del - 8p+dup - 8p22dup - 8q22.2del - 10p14del - 10p15.1del - 10p15.3del - 10q26.3del - 12pdup - 12p tetrasomy - 12p12.1del - 12p13.31del - 13qdel - 13q34del - 14q12del - Uniparental disomy of chrom. 14 - 15q11q13trip - 15q12 isodicentric - 15q13.3dup - 15q15q22del - 15q22.1q22.3del - 16p11.2p13.12trip - 16q12.1q13del - 16q12.2q21del - 16q24.3dup - 17p11.2del - 17q23.2q23.3del - 17q25dup - 18p11.2 to 1q42 unbalanced transl. - 19p12p11dup - 20p13p12.1dup - 21q22del - 21q22.3dup - 22 ring / r(22) - 22q11.21q11.23dup - Xp22.13dup - Xp22.33del - Xq26.3dup - Xq27q28dup -