Scientists unravel mystery of rare neurodevelopmental disorder, provide definitive diagnoses to 21 families worldwide

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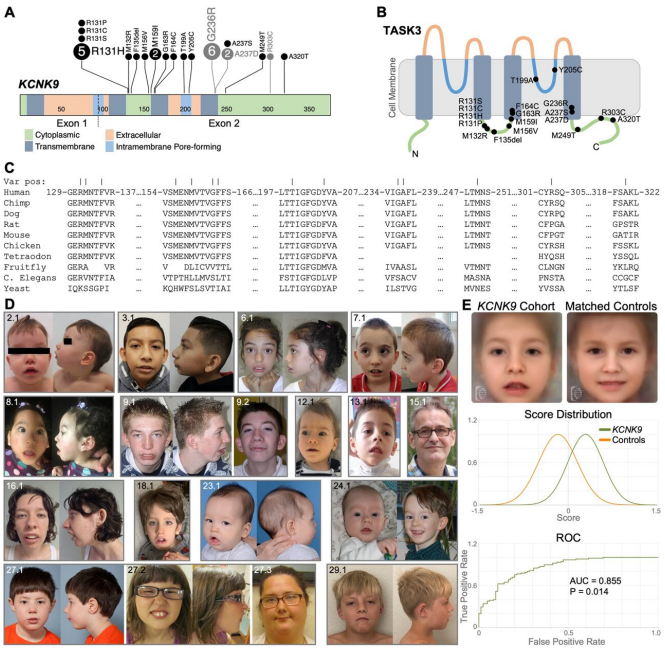

Individuals with *KCNK9* imprinting syndrome. **A** *KCNK9* coding exons showing all variants by predicted protein change using ProteinPaint (St. Jude Children's Research Hospital). The number in the circle represents the number of families described with the variant, no number = 1 family. Previously published variants are gray. **B** TASK3 protein topology schematic showing the location of variants. **C** Conservation of residues affected by variants. Bars in top line indicate variant position. **D** Front and profile photos of individuals with *KCNK9* variants. The age at time of photograph is: P2.1: 1y 6 m, P3.1: 9y, P6.1: 10y 3 m, P7.1: 9y, P8.1: 1y 5 m, P9.1: 12y-18y, P9.2: 12y-18y, P12.1: 2y 2 m; P13.1: 6y 1 m, P15.1: 58y, P16.1: 17y, P18.1 (updated photo, Pt. 1, Graham et al. 2016): 6y, P23.1: 1y 3 m, P24.1: left 3 m, right 5y, P27.1: 6y 1 m, P27.2: 8y1m, P27.3: 28y, P29.1: 8y 1 m. y = years; m = months. **E** Facial analysis using Face2Gene Research application (FDNA Inc. Boston, MA) of individuals with *KCNK9* imprinting syndrome (frontal photos of P3.1, P6.1, P7.1, P8.1, P9.1, P12.1, P13.1, P16.1, P18.1, P19.1*, P20.1*, P21.1*, 23.1, P24.1 (3 m photo), 25.1*, 27.1, and 29.1 compared to age, sex, and ethnicity matched controls. The aggregated binary comparison (AUC = area under the curve, ROC = receiver operating characteristic) demonstrates a significant difference between the two cohorts (*P*