08-1064: CITED2 and PBX1 in human adrenal development and disease

Supplemental Data: Clinical Characteristics

A. London, UK – CITED2 & PBX1 screened (n=36)

	46,XY (n=31)	**46,XX** (n=5)
Gonadal abnormalities e.g., microphallus, mild hypospadias, ambiguous genitalia and complete underandrogenization	13	-
Cardiac defects e.g., atrial septal defects, outflow tract anomalies	3	-
IUGR	5	-
Skeletal / IMAGe IUGR, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita and Genital Anomalies	5	1
Renal abnormalities e.g., glomerulosclerosis	2	-

Other features include brain abnormalities (1) and possible impaired glucose tolerance (1).
Number of patients previously screened for other adrenal candidate genes: NR5A1 (SF-1) = 35; NR0B1 (DAX1) = 35; StAR = 15; CYP11A1 = 16; MC2R (ACTHR) = 15; ACD = 5.

B. Dresden, Germany – PBX1 screened (n=20)

	46,XY (n=13)	**46,XX** (n=7)
Alacrima	5	1
Achalasia	3	3
Neurological abnormalities e.g., developmental delay, ataxia, epilepsy, hypotonia, nerve palsies	3	5
Muscle weakness	1	2
Hypogonadism	1	-

Other features include failure to thrive (2), diarrhea (2), respiratory infections / insufficiency (2), pancreatitis (1), cholelithiasis (1).
Number of patients previously screened for other adrenal candidate genes: NR0B1 (DAX1) = 2; MC2R (ACTHR) = 9; AAAS = 14.

C. Lyon, France – CITED2 screened (n=15)

	46,XY (n=8)	**46,XX** (n=7)
Deafness	-	1
IUGR / preterm	-	1
Neurological abnormalities e.g., developmental delay	1	-

Number of patients previously screened for other adrenal candidate genes: NR0B1 (DAX1) = 8 (males); MC2R (ACTHR) = 15; NR5A1 (SF-1) = 6; StAR = 5, CYP11A1 = 4.