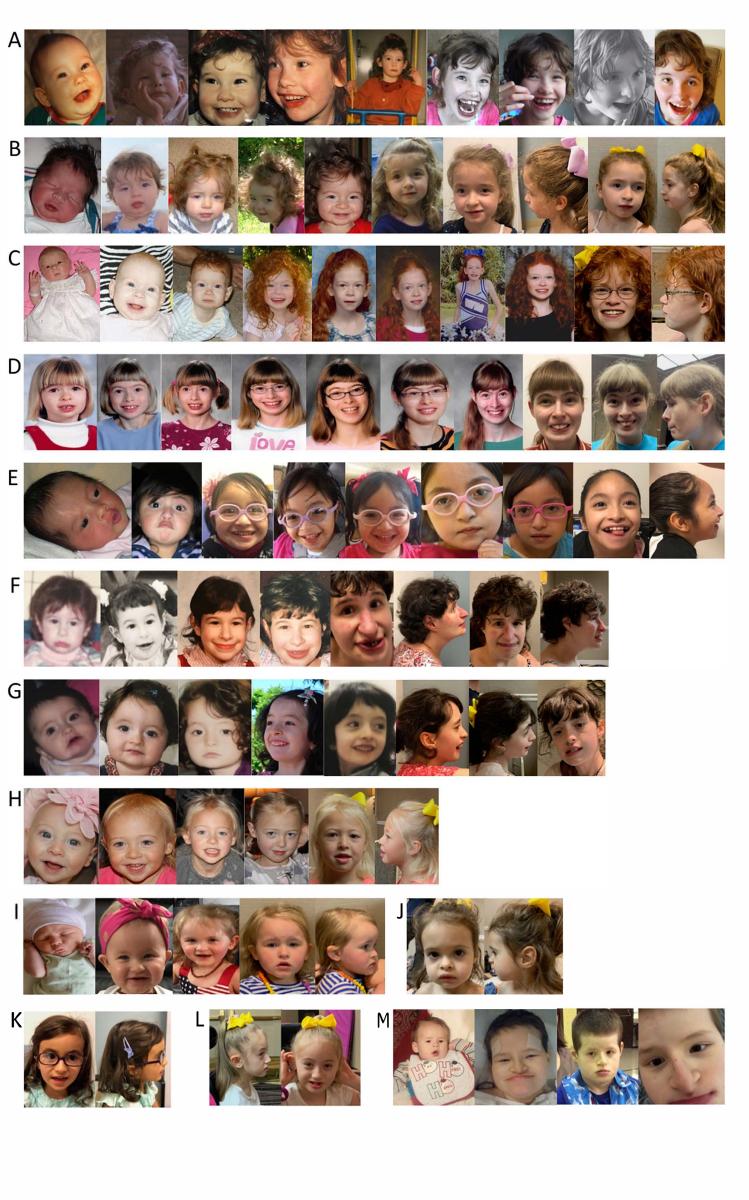
Subject ID	Inheritance	cDNA	Sex	Age	Alive Rason for NICU Stay	DD/ID			ASD, PDD- NOS, SCD, CDD, RETT	Autism Spectrum Disorder Testing	Social Communicatio n Questionnaire (SCO)	0, 12 0	PEDI-CAT Socia VCognitiv e Domain - T Score	VABSABC			VABS SOCIAL BASC - EXTERNALIZIN G T SCORE**	BASC - INTERNALIZIN G T SCORE	BASC - BEHAVIORAL SYMPTOMS INDEX T	BASC - ADAPTIVE SKILLS T SCORE
2	de novo		F	4	Y n/a	Y	Y	none	none ASD	N/A DSM-V .	n/c n/c	n/c n/c	n/c 12	47	n/c 38	48	n/c n/c 36 n/c	n/c n/c	n/c n/c	n/c n/c
3	de novo	c.616C>T	F	19	Y n/a	Y	N	none DAYC-2 (7 yo)	none	N/A	13	83	<10	28	20	20 :	38 52	50	61	19
4	de novo	c.616C>T	F	8	Y n/a	γ•	N	Cognifive: Age equiv. 13 months Receptive LanguAge: Age equiv. 12 months Expressive LanguAge: Age equiv. 10 months Gross Motor: Age equiv. 11 months Fine Motor: Age equiv. 12 months	ASD*	CARS2-ST (Age 7 yo) Total score 34.5	11	n/c	<10	n/c	n/c	n/c i	n/c n/c	n/c	n/c	n/c
								Goldman-Fristoe Test of Articulation, 2nd Edition (5y8m) Standard Score 86, 9% CAPUTE SCALES (Performed at 2y3m) LanguAge skills (CLAMS) = 10.3 month level Visual-motor/ problem-solving skills (CA) 3.3 month level		ADOS completed at 2y 3m but no concern for social disorder										
5		c.616C>T	F	3	Y Hypoglycemia, cyanosis Y n/a	Y*	N		none none	N/A	15	72 cannot score	<10 n/c	61 n/c	61 n/c			49 50	59 55	28 23
7	de novo de novo	c.616C>T	F	38	Y n/a Y n/a		N	none	PDD-NOS#	DSM-IV DSM-V .	17 14	79 n/c	<10 <10	n/c 31	n/c 20	n/c i		out of Age range 46	out of Age range 67	out of Age range 26
10	de novo	c.616C>T c.616C>T c.616C>T	F	18	Y n/a Y n/a		N		none	n/a n/a	14 24	n/c n/c	<10 22	n/c	n/c n/c	n/c i	n/c 51 n/c 64	42 64	59 70	26 27
11	de novo	c.616C>T	F	13	Y n/a	Y	N	none	none	n/a	n/c	n/c	<10	40	36	20 :	50 58	49	65	31
13	de novo	c.616C>T	F	27	Y n/a N n/a		N		none ASD	n/a none available	11 8	n/c n/c	n/c n/c		n/c n/c			46 n/c	58 n/c	26 n/c
14	de novo	c.616C>T c.616C>T c.616C>T c.616C>T c.616C>T c.616C>T	F	3	Y n/a		N		none ASD	n/a	20 14	81	37 <10		79 28			85 46	74 50	25 22
16	de novo	c.616C>T	F	17	Y n/a	Y**	· N	none VISC-4" (1 yo) Full scale (0 75 Varial comprehension 87 Perceptual Reasoning 79 Working Memory 68 Processing Speed 85 none	ASD	none available	28	n/c	<10	55	60	32	65 61 Out of Age	50 Out of Age	59	37 Out of Age
17	de novo	c.616C>T	F	23	Y n/a	Υ	Y	none	ASD	DSM-V	n/c	n/c	Range	62	63	57	62 Range	Range	Range	Range
18	de novo	c.616C>T	F	24	Y n/a	Y	N	none	none	n/a	23	70	Out of Age Range	25	30	20	22 n/c	n/c	n/c	n/c
40		Topic		20				School Aged neurodevelopmental assessment (8 yo 4mo) Peabody Test of Fine motor skills – 1st percentile Parent's Child Behavioural checklin- highly significant for dention and backline to the Behavioural checklin- highly significant for attention and though profile the control of the significant for attention and though ACTERS Questionnaire – significant for attention, hyperactivity, social skills Pediatric Developmental assessment (3 yo 8 mo) Bayley scales of infinat development. Mental scale and Stanford-Binet Intelligence Scale: Form L.M. – 18-22 month range; in keeping with significant delay (moderate mental handicas) Pediatric Developmental assessment clinic at 17 months Recoptive-Expressive Emergent Language Scale (REEL) showed moderate delay in integrals developments delay in integrals developments Bayley scale of Infinit development—overall 10.5-11.5 month Fine motor skills at 10-11 month level Gross motor skills at 11-12 month level	ASD#	DSM-IV criteria	18		Out of Age Range			25	Out of Age	Out of Age Range	Out of Age Range	Out of Age Range
20	de novo	c.616C>T c.616C>T c.616C>G	F	6	Y n/a		N		none	N/A	27	n/c	26		30	59	59 58	53	60	26
21	de novo	c.616C>G	F	20	Y n/a	Y	N		none	N/A	n/c	n/c	<10 Out of Age	ompl	20	20	20 n/c Out of Age	n/c Out of Age	n/c Out of Age	n/c Out of Age
		c.617G>A	F	36	Y n/a	Y	N	none Mullen Scales of Early Learning (3 yo) Visual Reception Scale T-score 20, 1st/k, Age Equiv. 11 mo, Very Low Fine Motor Scale T-score 20, 1st/k, Age Equiv. 15 mo, Very Low Receptive Language Scale T-score 20, 1st/k, Age Equiv. 16 mo, Very Low Receptive Language Scale T-score 20, 1st/k, Age Equiv. 10 mo, Very Low Early Learning Composite Scalandard Score 40, 415k, Very Low	none	N/A ADOS-2 Met Criteria for ASD (Module 1)	23	72	Range <10	28		33	37 Range	Range n/c	Range n/c	Range n/c
24	de novo	c.617G>A	М	17	Y Intubated for respiratory distres	3 Y	N	none	none	N/A	12	71	Out of Age	29	20	20	39 n/c	n/c	n/c	n/c
25	de novo	c.617G>A	F	17	Y Tachypneic	Υ	Y	none	ASD	CARS2-ST (Age 3 yo) Total score 52	16	n/c	n/c	n/c	n/c	n/c i	n/c 50	53	63	25
26	de novo	c.617G>A	F	11	Y n/a	Υ	N		Unk		17	n/c	n/c	n/c	n/c	n/c i		n/c	n/c	n/c
27	de novo	c.617G>T	F	36	Y n/a	γ.	· Y	LAPMER scale (Level of Activity in Profound/Severe Mental Retardation) (36yo): 4	PDD-NOS#	N/A	26	n/c	n/c	30	21	20	Out of Age 41 Range	Out of Age Range	Out of Age Range	Out of Age Range
28	de novo	c.617G>T c.626C>T c.629A>G c.634A>G c.635G>C	F	10	Y n/a Y meconium aspiration		N		none	N/A N/A	19 n/c	n/c n/c	<10 <10		n/c n/c		n/c 46	48 n/c	60 n/c	20 n/c
30	de novo	c.634A>G	F	5	Y n/a	Y	Y	none	CDD*	none	16	72	n/c	60	54	55	67 n/c	n/c	n/c	n/c
		c.638C>T		20	Y n/a	Y	N	none WISC-V (1 yo) Full Scale (0 75 Verbal Congrehension 87 Perceptual Reasoning 79 Working Memory 88 Processing Speed 85	PDD-NOS #, SCD	N/A ASDS (11 yo) Asperger's Syndrome Quotient 107, 68th% (likely Asperger's Syndrome)	12 n/c	74	<10	71	75	89	Out of Age	39 Out of Age Range	59 Out of Age Range	33 Out of Age Range
								DAYC-2 (Ape 44/2014) Cognitive (SS-Y2) Social Emotionia (SS-Y5) Physical Developmental (SS-98) Constant Developmental (SS-98) Physical Developmental (SS-98) Constant Developmental (SS-98) Battelle Developmental Inventory, Second Edition (4y8m) Attention and Memory, Scaled Score 1, SS-95, 4-1%, Entermely low Perception and Concepte Scaled Score 1, SS-95, 4-1%, Entermely low Adaptive Behavior Assessment System-Second Edition (ABAS-8) (4y8m), Total Concepts Scaled Score 1, SS-95, 4-1%, Entermely low Adaptive Behavior Assessment System-Second Edition (ABAS-8) (4y8m), Including Social-Emotional Domain) Preschool Language Sociale, Fifth Edition (PS-5) (4y8m) Auditory Comprehension: SS-950 Expressive Communication: SS-950 Total Language Score: SS-950 CAPUTE SCALE-S6 (4y) Language skills (CLAMS) = 9.3 month level, DO-19 Valual-motory problem-solving skills (CAT) 12.1 month level, DO-2 5		ADOS- 2nd Ed. Resulbi: Social Affect (Communication & Reciprocal Social Interaction): 17 Restricted and Repetitive Behavior: 8 Total Score: 25 Comparison Score (Level of audism spectrum-related symptoms; 9: high level of audism spectrum-related symptoms.										are gr
33	ae novo	c.1019A>T	М	8	Y n/a	Y*	N		ASD*		14	n/c	<10	n/c	n/c	n/c i	n/c n/c	n/c	n/c	n/c

Supplemental Table 1: Additional Testing including Cognitive Scores, Autism Spectrum Disorder testing N/A = not applicable, n/c = notcompleted, none = not done, ADOS = Autism Diagnostic Observation Schedule, CARS2-ST = Childhood Autism Rating Scale, 2nd Edition, Standard Version, DAYC-2 = Developmental Assessment of Young Children, 2nd Edition, DQ = developmental quotient, SCD = Social Communication Disorder, SS = standard score, WISC-IV = Wechsler Intelligence Scale for Children, Fourth Edition. # Denotes parent reports diagnosis















Supplemental Figure 1: Sequential Facial Pictures of Individuals Carrying Variants in *HNRNPH2*

Participant 1 at 1 year, 2 years, 3 years, 5 years, 7 years, 10 years, 16 years, 24 years, and 24 years (A); Participant 2 at 1 month, 1 year, 2 years, 3 years, 4 years, 5 years, 7 years, 7 years, 8 years, and 8 years (B*); Participant 3 at 2 days, 5 months, 1 year, 5 years, 7 years, 9 years, 13 years, 16 years, 17 years, and 17 years (C); Participant 4 at 3.5 years, 5.5 years, 9 years, 11 years, 13 years, 15 years, 17 years, 21 years, 22 years, and 22 years (D); Participant 5 at 6 months, 2 years, 3 years, 4 years, 5 years, 6 years, 8 years, 10 years, 10 years (E); Participant 6 at 9 months, 3 years, 4 years, 8 years, 37 years, 37 years, 38 years, and 38 years (F*); Participant 7 at 6 months, 16 months, 2 years, 3 years, 4 years, 11 years, 12 years, and 12 years (G); Participant 8 at 1.5 years, 2 years, 4 years, 5 years, 5 years, and 5 years (H); Participant 9 at 3 days, 1 year, 2 years, 3 years, and 3 years (I); Participant 10 at 3 years and 3 years (J); Participant 11 at 17 years and 17 years (K); Participant 13 at 7 years and 7 years (L); Participant 14 at 2 years, 13 years, 13 years, and 16 years (M); Participant 15 at birth, 2 months, 1 year, 18 months, 6 years, 10 years, 16 years, 19 years, and 18 years (N); Participant 16 at 1 months, 3 months, 3 months, 1 year, 1 year, 1 year, 2 years, 2 years, and 2 years (O); Participant 17 at 1 year, 2 years, 5 years, and 8 years (P); Participant 18 at 1 year, 2 years, 4 years, 6 years, 8 years, 14 years, and 16 years (Q^); Participant 19 at 2 years, 4 years, 18 years, 23 years, 26 years, 36 years, and 36 years (R); Participant 20 at 2 days, 8 months, 3 years, and 4 years (S); Participant 21 at 1 year, 3 years, 5 years, 6 years, 7 years, and 7 years (T); Participant 22 at 3 years, 5 years, 6 years, 7 years, 10 years, 19 years, 20 years, and 20 years (U); Participant 23 at 1 month, 3 months, 6 months, 1 year, 4 years, 5 years, 6 years, 7 years, 8 years, and 9 years (V). * Denotes previously described in Bain et al, 2016 and ^ denotes described in Peron et al 2020.