



## Anatomical Location of non-ectopic supernumerary nipples (Embryonic Milk line)

- Additional Mammary glands & nipples develop from embryonic Milk line (a ventral epidermal ridge) extending from axilla to groin; which result from failure of regression during human embryogenesis.
- Atavism: presence of evolutionary regressed ancestor trait

## Insight:

- 1. What are the key characteristic differences between SGBS type 1 & type 2?
- 2. What is the meaning of 'positional cloning' & 'overgrowth syndrome'?
- 3. Is there any genetic heterogenicity for SGBS?
- 4. How would you counsel III:2 & III:3 for case IV:3?
- 5. What are the surveillance guidelines for an overgrowth syndrome?

	Plausible tenets:		S
	Gene: GPC3 (Xq26.2); 451,356 bps & 8 Exons		- Sy IN
	- Glypican's belongs to the glypican-related integral membrane		D
	- Transcript length: 2267 bps, eight domains & features, thirteen		F
1	splice variants, 205 orthologues, five paralogue		H
X.	- Proteoglycan Protein of cell surface [(580 AA), Molecular mass:		
3	65563 Da] carries heparan sulfate.		B S
	- Negatively regulator for hedgehog signaling(SHH) pathway by		
	opposing for receptor while positively regulates the Wnt signaling		S
A.	patnway & do fine-tuning of BMP4, BMP2, BMP7, & FGF7 proteins		P D
2	- Might have a role in growth regulation & cell division		sy
	Clinical Phenotypes		D E
4	- X Linked inheritance pattern (XLR)		E D
	- No clincial diagnositic criteria		C.
4	- <b>Type 1 SGBS</b> : Primordial overgrowth syndrome(OGS),		
1.14	higher cognitive issues		IN D
AR	- <b>Other anomalies reported:</b> coarse facial features.		W
	supernumerary nipples, focal ectodermal defect(abdominal &		II
	diaphragmatic hernia), skeletal dysplasia ( segmented vertebral		D
	defect & postaxial polydactyly), high risk for embryonal tumors (		D
	As ominous for the majority of OGS)		B
	- Management: symptomatic with Multidisciplinary approach		3
X	- Investigation. Johow the protocol used for any dyshorphic case(https://www.nchi.nlm.nih.gov/nmc/articles/PMC4324078/)		C
	- Serum alpha fetoprotein (5-10 % risk for embryonic tumor)		D
2			A
ALL C		┥	E C
A.S.	<b>Type 2 SGBS (XLR):</b> Gene <b>OFD</b> , brachydactyly, recurrent respiratory infection ( <b>ciliary</b>		_
	<b>Genetic heterogeneity with type 1 SGBS: 30 %</b> SGBS fail to spot by GPC3/4 testing		P/ SY
		٦	12
Z	Resistional classing (around the 1000c), leasting of a cone (unly come for sting)		N D
	activity) position in the genome by known polymorphic markers in a family having		H L
4	multiple affected cases.		D
20		5	A
3	<b>Overgrowth syndrome:</b> should be suspected if any two anthropometric parameters-	\$	C/ 0
	head circumferences, height, & weight are > 2 SD from the mean		SY
B	Imperative Counselling aspects for case IV-3		F/
	- Confirming the diagnosis: case IV:2, & IV:4 (in view positive of consanguinity need to		A R
	rule inherited haemoglobinopathy especially thalassemia)		E
	- consanguinity could increase the risk of all types of mendelian MOI		H
	Thought Rivoting		A C
	How do the shelestered levels interact with CDC2 at the sellular level	Ľ	IN
No.	How do the cholesterol levels interact with GPCS at the centular level		D
	beyond the embryonal period?	0	CI
	Can CEmRNA (Carcinoembryonic microRNA panel) be used as the		
-	earliest biochemical marker for embryonal tumors?	A	
J.	What is the relation of GPC3 protein to the cellular aging process (CAP)?		т
	How does the duplication of exons 1-9 in GPC4 alter GPC3 expression?	III	1
	Which alternative splice variants of CDC2 have an active role in the	-	0
4	which alternative splice variants of GPUS have an active role in the		
	regression of the embryonic milk line?	E B	M
1	Does a genetic program or pathway regress or stimulate atavism & how		S
	does it help in evolution?	ALC: N	C
		1	1
		and the	52

John Strephysics     OGT (XLR)     Distribution       DEVELOPMENT DISORDER X linked 106; XLI D106     OGT (XLR)     D + MR changes & microcephaly       DOCAL DERMAL     PORCN     Kocal skin changes & microcephaly       NICONTINENTIA PIGMENTI     IKBKG     Classical skin changes & microcephaly       SYNDROME     TFAP2A     Branchial cleft sinus clefts, ocular anomalies, conductive hearing loss of culoecroderential dermoid & gyndrome)       OCULOECTODERMAL SYNDROME     KRAS     Epibulbar dermoid & aplasia cutis congenita       OSTAXIAL ACROFACIAL DYDSOTOSIS (Miller     DHODH     Severe micrognathia, coloboma of eyelids, uhar ray defects       DEVELOPMENTAL AND DEVELOPMENTAL AND DEVELOPMENTAL AND DEVELOPMENT DISORDER     MIDH2 (AR)     Multisuture cranicosynostosis, polysyndactyly & obesity       INTELLECTUAL DEVELOPMENT DISORDER     DDX6 (AD)     Monormal philtrum, cradiac & urogenital anomales & friendly behavior       TRICHOODONTOONYCHIAL DYSPLASIA WITH BONE DEFICIENCY     ? (AR)     Ectodermal dysplasia, nevus pigmentosus, frontoparietal bone deficiency       BOHRING-OPTTZ SYNDROME; BOPS     ASXL1     Severe IUGR, trigonocephaly, nevus flammeus, forehead hirsutism       CHARCOT-MARIE-TOOTH DISASE, TYPE 4A; CMT4A     GDAP1 (AR)     Streaks of hypo- or hypertelorism, short neck       NURVLOBLEPHARON ECTODERMAL DEFECTS- UALOSUM, CA	Syndromes with sug	pernumer Gene (MOI)	ary nipples
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AND DYSMORPHIC FACIES; IDDILF TRICHOODONTOONYCHIAL DYSPLASIA WITH BONE DEFICIENCY BOHRING-OPITZ SYNDROME; BOPS ASXL1 CHARCOT-MARIE-TOOTH DISEASE, TYPE 4A; CMT4A ANKYLOBLEPHARON ECTODERMAL DEFECTS- CLEFT LIP/PALATE; AEC PALLISTER KILLIAN SYNDROME Tetrasomy of 12p DISORDER WITH HYPOTONIA, IMPAIRED LANGUACE, AND DISORDER WITH HYPOTONIA, IMPAIRED LANGUACE, AND DYSMORPHIC FEATURES; NEDHILD SYNDROME; ACOGS BLEPHAROPHICOSIS WITH ACLAL AND GENITAL SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACLAL AND GENITAL SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACLAL AND GENITAL SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACLAL AND GENITAL ANDALES AND MENTAL DYSMORPHIC FEATURES; NEDHILD SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACLAL AND GENITAL ANDALES AND MENTAL SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACLAL AND GENITAL ANDALES AND MENTAL SYNDROME; ACOGS BLEPHAROPHINOSIS WITH ACALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGS BLEPHAROPHIMOSIS WITH ACIAL AND GENITAL ANDALIES AND MENTAL SYNDROME; ACOGS BLEPHAROPHIMOSIS WITH ACALLOSUM HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUM HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUMAL HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUMAL HYPOTHYROIDISMAND AGENESIS OF THE	WITH IMPAIRED LANGUAGE		cardiac & urogenital
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CLEFT LIP/PALATE; AEC   Synechiae, maxinary     PALLISTER KILLIAN   (Somatic mosaicism)     SYNDROME Tetrasomy of 12p   Streaks of hypo- or hyper-pigmentation, hypertelorism, short neck     NEURODEVELOPMENTAL   CHAMP1     DISORDER WITH   (AD)     HYPOTONIA, IMPAIRED   CHAMP1     LANGUAGE, AND   Ophthalmologic anomalies, sleep disturbances, seizures     DYSMORPHIC FEATURES;   Desterment     NEDHILD   CDH2 (AD)     AGENESIS OF CORPUS   CDH2 (AD)     OCULAR AND GENITAL   CMAC (AGG)     SYNDROME; ACOGS   CDH2 (AD)     BLEPHAROPHIMOSIS WITH   ?(AR)     FACIAL AND GENITAL   ?(AR)     Midface hypoplasia, ear anomalies, unanomalies,	ANKYLOBLEPHARON	TP63 (AD)	Eyelids & alveolar
PALLISTER KILLIAN SYNDROME Tetrasomy of 12p(Somatic mosaicism)Streaks of hypo- or hyper-pigmentation, hypertelorism, short neckNEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILDCHAMP1 (AD)Ophthalmologic anomalies, sleep disturbances, seizuresNEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILDCHAMP1 (AD)Ophthalmologic anomalies, sleep disturbances, seizuresAGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGSCDH2 (AD)ID, behavioral abnormalities, downslanting palpebral fissures, HyposmiaBLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION?(AR)Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephaly ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUM?(? AR/XL/ contiguous gene syndrome)Ectopic or absent thyroid, Severe ID, midface hypoplasia, macrocephaly, hypotoniaINTELLECTUAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65KDM5B (AR) AM of 5th finger CamptodactylyDownslanting palpebral fissures, ear anomalies, thin lips, 4th or 5th finger CamptodactylyCHAR SYNDROMETFAP2B (AD)Downslanting palpebral fissures, thick lips, hypertelorism, mild ID, or the torism, mild ID, or the torism, mild ID,	CLEFT LIP/PALATE; AEC		synechiae, maxiliary
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12p   hypertelorism, short neck     NEURODEVELOPMENTAL   CHAMP1     DISORDER WITH   (AD)     HYPOTONIA, IMPAIRED   (AD)     LANGUAGE, AND   DYSMORPHIC FEATURES;     NEDHILD   CDH2 (AD)     AGENESIS OF CORPUS   CDH2 (AD)     CALLOSUM, CARDIAC,   CDH2 (AD)     OCULAR AND GENITAL   abnormalities,     SYNDROME; ACOGS   fissures, Hyposmia     BLEPHAROPHIMOSIS WITH   ?(AR)     FACIAL AND GENITAL   Widface hypoplasia, ear     ANOMALIES AND MENTAL   Underdeveloped     RETARDATION   gene     ECTODERMAL DYSPLASIA,   ?(? AR/XL/     COTDERMAL DYSPLASIA,   ?(? AR/XL/     ECTODERMAL DYSPLASIA,   ?(? AR/XL/     MYPOHIDROTIC, WITH   contiguous     HYPOHIDROTIC, WITH   contiguous     HYPOTHYROIDISM AND   gene     AGENESIS OF THE CORPUS   syndrome)     CALLOSUM   midface hypoplasia,     INTELLECTUAL   KDM5B   Downslanting palpebral     INTELLECTUAL   KDM5B   Downslanting palpebral     DISORDER, AUTOSOMAL   TFAP2B   Downslanting palpebr	SYNDROME Tetrasomy of	mosaicism)	hyper-pigmentation,
NEURODEVELOPMENTAL DISORDER WITH CHAMP1 (AD) Ophthalmologic anomalies, sleep disturbances, seizures   NEURODEVELOPMENTAL DISORDER WITH (AD) Ophthalmologic anomalies, sleep disturbances, seizures   NEURODEVELOPMENTAL LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILD CDH2 (AD) ID, behavioral abnormalities, downslanting palpebral fissures, Hyposmia   AGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGS CDH2 (AD) ID, behavioral abnormalities, downslanting palpebral fissures, Hyposmia   BLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION ?(AR) Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephaly   ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUM ?(? AR/ XL/ contiguous gene syndrome) Ectopic or absent thyroid, Severe ID, midface hypoplasia, macrocephaly, hypotonia   INTELLECTUAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 KDM5B (AR) Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	12p		hypertelorism, short
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HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILDdisturbances, seizuresAGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGSCDH2 (AD)ID, behavioral abnormalities, downslanting palpebral fissures, HyposmiaBLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION?(AR)Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephalyECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH HYPOTHYROIDISM AND AGENESIS OF THE CORPUS 	DISORDER WITH	(AD)	anomalies, sleep
LANGOAGE, AND DYSMORPHIC FEATURES; NEDHILD AGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGS BLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION CTODERMAL DYSPLASIA, HYPOHINOTIC, WITH HYPOHINOTIC, WITH HYPOHINOTIC, WITH HYPOHINOTIC, WITH HYPOHINOTIC, WITH CALLOSUM AGENESIS OF THE CORPUS CALLOSUM INTELLECTUAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL DEVELOPMENTAL CALLOSUM INTELLECTUAL DEVELOPMENTAL DEVELOPMENTAL CALLOSUM CHAR SYNDROME TFAP2B (AD) HYPOTHYROIDIS, MID CHAR SYNDROME TFAP2B (AD) HYPOTHYRO PATION CALLOSUM CHAR SYNDROME CHAR SYNDROME CHAR SYNDROME CALCON CALCON CALCON CALCON CALCON CHAR SYNDROME CALCON CA	HYPOTONIA, IMPAIRED		disturbances, seizures
NEDHILD     AGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL   CDH2 (AD)   ID, behavioral abnormalities, downslanting palpebral fissures, Hyposmia     BLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION   ?(AR)   Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephaly     ECTODERMAL DYSPLASIA, HYPOHINOTIC, WITH HYPOHINOTIC, WITH HYPOHINOTIC, WITH CALLOSUM   ?(? AR/XL/ contiguous gene syndrome)   Ectopic or absent thyroid, Severe ID, midface hypoplasia, macrocephaly, hypotonia     INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65   KDM5B (AR)   Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly     CHAR SYNDROME   TFAP2B (AD)   Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	DYSMORPHIC FEATURES:		
AGENESIS OF CORPUS CALLOSUM, CARDIAC, OCULAR AND GENITAL SYNDROME; ACOGSCDH2 (AD) abnormalities, downslanting palpebral fissures, HyposmiaBLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION?(AR)Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephalyECTODERMAL DYSPLASIA, HYPOHINDOTIC, WITH HYPOHTHROIDISM AND AGENESIS OF THE CORPUS CALLOSUM?(? AR/XL/ contiguous gene syndrome)Ectopic or absent midface hypoplasia, macrocephaly, hypotoniaINTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65KDM5B (AR)Downslanting palpebral fissures, ear anomalies, thin lips, 4th or 5th finger CamptodactylyCHAR SYNDROMETFAP2B (AD)Downslanting palpebral fissures, thick lips, hypertelorism, mild ID, or with the lips, 4th or 5th finger	NEDHILD		
CALLOSUR, CADIAC, abnormalities,   OCULAR AND GENITAL downslanting palpebral   SYNDROME; ACOGS fissures, Hyposmia   BLEPHAROPHIMOSIS WITH ?(AR) Midface hypoplasia, ear   FACIAL AND GENITAL ?(AR) Underdeveloped   ANOMALIES AND MENTAL Underdeveloped   RETARDATION ?(? AR/XL/ Ectopic or absent   HYPOHIDOTIC, WITH contiguous thyroid, Severe ID,   HYPOHTHROIDISM AND gene midface hypoplasia,   AGENESIS OF THE CORPUS syndrome) macrocephaly,   INTELLECTUAL KDM5B Downslanting palpebral   DISORDER, AUTOSOMAL (AR) fissures, ear anomalies,   RECESSIVE 65; MRT65 TFAP2B Downslanting palpebral   (AD) fissures, thick lips, hypertelorism, mild ID,	AGENESIS OF CORPUS	CDH2 (AD)	ID, behavioral
SYNDROME; ACOGS downstanting papebrail   BLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION ?(AR) Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephaly   ECTODERMAL DYSPLASIA, HYPOHIDOTIC, WITH HYPOHIDROTIC, WITH CALLOSUM ?(? AR/XL/ contiguous Ectopic or absent thyroid, Severe ID, midface hypoplasia, gene syndrome)   INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 KDM5B (AR) Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	OCULAR AND GENITAL		abnormalities, downslanting palpohral
BLEPHAROPHIMOSIS WITH FACIAL AND GENITAL ANOMALIES AND MENTAL RETARDATION ?(AR) Midface hypoplasia, ear anomalies, Underdeveloped genitals, trigonocephaly   ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH HYPOHIDROTIC, WITH GALLOSUM ?(? AR/XL/ contiguous gene syndrome) Ectopic or absent thyroid, Severe ID, midface hypoplasia, gene syndrome)   INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 KDM5B (AR) Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	SYNDROME; ACOGS		fissures. Hyposmia
FACIAL AND GENITAL   anomalies,     ANOMALIES AND MENTAL   underdeveloped     RETARDATION   genitals, trigonocephaly     ECTODERMAL DYSPLASIA,   ?(? AR/XL/     HYPOHIDOTIC, WITH   contiguous     HYPOHIDOTIC, WITH   gene     AGENESIS OF THE CORPUS   syndrome)     CALLOSUM   KDM5B     INTELLECTUAL   KDM5B     DEVELOPMENTAL   (AR)     DISORDER, AUTOSOMAL   (AR)     RECESSIVE 65; MRT65   TFAP2B     CHAR SYNDROME   TFAP2B     (AD)   bypertelorism, mild ID,	BLEPHAROPHIMOSIS WITH	?(AR)	Midface hypoplasia, ear
ANUMALES AND MENTAL RETARDATION Underdeveloped genitals, trigonocephaly ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH HYPOHIDROTIC, WITH HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUM NOT CALLOSUM INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 CHAR SYNDROME TFAP2B (AD) FOR A CALLOSUM INTELLECTUAL CALLOSUM CHAR SYNDROME TFAP2B (AD) CALLOSUM INTELLECTUAL CALLOSUM CHAR SYNDROME TFAP2B (AD) CALLOSUM INTELLECTUAL CALLOSUM CHAR SYNDROME TFAP2B (AD) CALLOSUM CHAR SYNDROME CALLOSUM CHAR SYNDRO	FACIAL AND GENITAL		anomalies,
ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH ?(? AR/XL/ contiguous Ectopic or absent   HYPOHIDROTIC, WITH contiguous thyroid, Severe ID, midface hypoplasia, macrocephaly, hypotonia   AGENESIS OF THE CORPUS CALLOSUM syndrome) midface hypoplasia, macrocephaly, hypotonia   INTELLECTUAL KDM5B Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   DISORDER, AUTOSOMAL TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	ANUMALIES AND MENTAL RETARDATION		Underdeveloped
HYPOHIDADTIC, WITH HYPOHIDADTIC, WITH AGENESIS OF THE CORPUS CALLOSUM contiguous gene syndrome) thyroid, Severe ID, midface hypoplasia, macrocephaly, hypotonia   INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 KDM5B (AR) Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	ECTODERMAL DVSPI ASIA	?(? AR / YI /	genitals, trigonocephaly Ectopic or absent
HYPOTHYROIDISM AND AGENESIS OF THE CORPUS CALLOSUM gene syndrome) midface hypoplasia, macrocephaly, hypotonia   INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 KDM5B (AR) Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	HYPOHIDROTIC, WITH	contiguous	thyroid, Severe ID.
AGENESIS OF THE CORPUS CALLOSUM syndrome) macrocephaly, hypotonia   INTELLECTUAL KDM5B Downslanting palpebral fissures, ear anomalies, thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	HYPOTHYROIDISM AND	gene	midface hypoplasia,
KDMSB     hypotonia       INTELLECTUAL     KDMSB     Downslanting palpebral       DEVELOPMENTAL     (AR)     fissures, ear anomalies,       DISORDER, AUTOSOMAL     thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger       RECESSIVE 65; MRT65     Camptodactyly       CHAR SYNDROME     TFAP2B     Downslanting palpebral       (AD)     fissures, thick lips, hypertelorism, mild ID,	AGENESIS OF THE CORPUS	syndrome)	macrocephaly,
INTELECTION KDM5B Downstanting paipebral   DEVELOPMENTAL (AR) fissures, ear anomalies, thin lips, 4th or 5th finger   DISORDER, AUTOSOMAL Camptodactyly   CHAR SYNDROME TFAP2B Downslanting palpebral   (AD) fissures, thick lips, hypertelorism, mild ID,		KDMED	hypotonia
DISORDER, AUTOSOMAL RECESSIVE 65; MRT65 thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger Camptodactyly   CHAR SYNDROME TFAP2B (AD) Downslanting palpebral fissures, thick lips, hypertelorism, mild ID,	INTELLECTUAL DEVELOPMENTAL	(AR)	fissures ear anomalies
RECESSIVE 65; MRT65 CHAR SYNDROME (AD) CHAR SYNDROME (CHAR SYNDROME (CHAR SYNDROME (CHAR SYNDROME) CHAR SYNDROME) CHAR SYNDROME (CHAR SYNDROME) CHAR SYNDROME (CHAR SYNDROME) CHAR SYNDROME (CHAR SYNDROME) CHAR SYNDROME (CHAR SYNDROME) CHAR SYNDROME (CHAR SYNDROME)	DISORDER, AUTOSOMAL	(int)	thin lips, 4 <sup>th</sup> or 5 <sup>th</sup> finger
CHAR SYNDROME TFAP2B Downslanting palpebral (AD) fissures, thick lips, hypertelorism, mild ID,	RECESSIVE 65; MRT65		Camptodactyly
(AD) fissures, thick lips, hypertelorism, mild ID,	CHAR SYNDROME	TFAP2B	Downslanting palpebral
hypertelorism, mild ID,		(AD)	fissures, thick lips,
Vioonwallring			nypertelorism, mild ID, Sleenwalking
Parasomnia			Parasomnia
TYSHCHENKO SYNDROME ? (AD) Short stature, flattened	TYSHCHENKO SYNDROME	? (AD)	Short stature, flattened
face, Prominent eye,			face, Prominent eye,
CLIVER SYNDROME 2(AD) Destruction of the second	OLIVED CVNDDOME	2(4.0)	congenital heart disease
VERVER STRUKOWE (AK) POSTAXIAI POLYdactyly, svndactyly, nrominent	OLIVER STINDRUME	(AK)	syndactyly, prominent
jaw & eyebrows, ID			jaw & eyebrows, ID

pastic gait, pyramidal syndrome & behavioral disorders, CP-erebral palsy.

uthors: Dr shant Ku vati -