chromosome 10p and monosomy 15q26.3- >qter which contains the region coding for IGF1R. IGF-1(243 μg/L) and IGFBP-3 (3501 ng/ml) levels were markedly elevated; +4.7 SDS and +3.3 SDS respectively. The marked elevation in IGF-1 levels was considered a relative contraindication to GH therapy.

Discussion Haploinsufficency of IGF1R gene is associated with elevated levels of IGF-1 as a result of target organ resistance. Exogenous growth hormone, while further elevating IGF-1, in many cases facilitates catch-up growth albeit to a lesser extent than other ex-SGA infants. In this case IGF-1 levels were exceptionally high, precluding growth hormone therapy.

Conclusion Deletions involving the IGF1R gene are a rare but treatable cause of short stature. This case is unusual however, as marked elevation of IGF-1 at baseline precluded GH therapy.

PO-0070 PECULIARITIES OF THE GROWTH HORMONE AND INSULIN-LIKE GROWTH FACTOR (IGF-1, IGFBP-3) SECRETION IN GENETICALLY DETERMINED TYPES OF SHORT STATURE IN UZBEKISTAN

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Goal To study the growth hormone (GH) and insulin-like growth factor (IGF-1, IGFBP-3) secretion in genetically determined types of short stature (GDSS) in Uzbekistan.

Materials and methods We examined 92 patients with GDSS [11 - Russel-Silver syndrome (RSS), 16 - Noonan syndrome (NS), 10 - Sekkel syndrome (SS), 9 - Prader-Willi syndrome (PWS), 8 - Cornelius de Lange syndrome (CLS), 38 girls with TS] at the age of 7–18. A level of GH, IGF-1, IGFBP-3, anthropometry (SDS) was studied.

Results Stunting of various degrees of expression was observed in all patients with GDSS but it was most expressed in patients with RSS (-5.16 ± 1.18 SDS), SS (-4.18 ± 1.12 SDS) and CLS (-6.10 ± 1.14 SDS). A reliably low level of GH vs. the control was found in patients with CLS (0.64 ± 0.05 ng/ml, $p < 0.05$), RSS (0.7 ± 0.04 ng/ml, $p < 0.05$), SS (1.02 ± 0.07 ng/ml, $p < 0.05$) on the background of a low level of IGF-1 and IGFBP-3. Patients with NS, TS and PWS had a level of GH within the lower limit norm, 12 girls with TS had a GH level which was reliably low (0.04 ± 0.05 ng/ml, $p < 0.05$) but IGF-1 and IGFBP-3 rates corresponded to the lower limit of the age norm.

Conclusions In Uzbek patients with GDSS pronounced stunting was found in patients with RSS, SS and CLS which was associated with disturbances of a direct and reverse relation in the GH-IGF-IGFBP-3 system. A low GH level and relative deficiency of IGF-1 and IGFBP-3 in girls with TS was related with disturbances in the pituitary-ovarian system.

PO-0071 ROLE OF GROWTH HORMONE THERAPY IN CLINICAL, LABORATORY, RADIOLOGICAL IMPROVEMENT OF CHILDREN WITH VITAMIN-D DEPENDENT RICKETS TYPE-2

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Rickets is a disease in on growing bones due to precipitate disorders of calcium (Ca) and phosphor (P) in matrix of growth plate before epiphysial closing. Different types for rashitism including nutritional, hypophosphatemic and vitamin-D dependent rickets) VDDR). VDDR is a result of severe vitamin D deficiency that had tow subtype:

A) type -1(VDDR-1) because of defect of 1-alfa-hydroxilase enzyme.

B) type -2(VDDR-2) because of mutation in intracellular vitamin-D receptor (VDR) associated with some especial symptoms like alopesia (global head hair loss) and short stature. this disease needs to high dose of vitamin-D analogues also calcium complement. although response to treatment is variable.

Cases Introduction We introduce 3 cases of VDDR-2 in a family (2 sisters aged 1.5, 5 years and their 5 years old cousin) who referred to endocrine and metabolic centre in Emam–Reza hospital in Mashhad presenting limb deformities, wide wrist, seizure, disorder in dental growth, alopesia, several bone fractures. they had not been recovered despite receiving sufficient treatment. it was notable when they received growth hormone therapy because of growth failure, their rickets improved dramatically by clinical, laboratory, radiological.

Discussion According to discover several clinical indication for growth hormone therapy during last 50 years, still there are not enough studies about growth hormone therapy in VDDR. As second type of vitamin-D dependent rickets sometimes needs to long time calcium infusion or high dosage of active vitamin-D analogues, so it seems that growth hormone therapy in these patients especially with short stature may improve linear growth and rickets symptoms too.

PO-0072 A BOY WITH DECELERATED LINEAR GROWTH WITH NORMAL GROWTH HORMONE (GH) – INSULIN-LIKE GROWTH FACTOR–I (IGF-I) AXIS HAD AN EXCEPTIONAL RESPONSE TO GH THERAPY

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Y M is a boy who presented at the age of 5.5 years with slow growth He had a birth weight = 3.1 kg and length = 49.5 cm. Infantile and early neonatal periods are uneventful. He had normal development and appropriate nutritional history. No family history of endocrinopathy and/ or short stature was reported. His measurements were: weight= 16.9 kg, Height = 104 cm (HtSDS = -1.8), growth velocity (GV = 1 cm/y) and BMI= 16 and mid-parental height (MPHt) SDS = -0.6. He had no

Abstract PO-0072 Table 1 Growth data of the patient before and after GH therapy

Age /year	BMI	HSDS	IGF1 ng/ml	Bone age years	Notes Tanner stage
4.48	16	-0.4	65	ND	
5.5	16	-1.84	70	3	Started GH
5.7	16	-0.7	92		
6.4	16	-0.35	148	4	
7	15	-0.28	279		
8	17	0.15	295		
9	18	0.5	195	8.5	
10	20	0.5	325		Tanner 1
11	22	0.5	447		Tanner 2
12	22	0.83	514	12.5	Tanner 3

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