GP72 OBESITY RELATED GLOMERULOPATHY IN THREE SIBLINGS

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Introduction Obesity is an urging health care problem with its increasing incidence in years. Renal involvement secondary to obesity is called obesity related glomerulopathy (ORG). ORG is characterized with heavy proteinuria without evident edema, and progressive renal dysfunction. Histological features include focal segmental glomerulosclerosis (FSGS), mesangial expansion, glomerulomegaly and <50% foot process effacement by electron microscopy. We hereby present 3 siblings with ORG.

Case report Proteinuria was first detected in a 12.5 year-old female patient, after a urinary tract infection. Further investigation revealed proteinuria in her 2 other siblings aged 16.5 and 7.5. All three of them were obese females and they had no prominent edema (table 1). Two of them had HT and nephrotic range proteinuria; and their renal biopsies revealed FSGS. In the other sibling, biopsy was not diagnostic. The oldest sibling's biopsy also showed chronic parenchymal injury signs. Family history revealed that their father had renal transplantation secondary to renal failure with unknown etiology at the age of 20. Moreover they also had one cousin with renal failure requiring hemodialysis. Due to their heavy family history and consanguinity of the parents, genetic testing was made; but we couldn't find any identified causative mutations. All three of them were started on enapril which is an angiotensin-converting enzyme (ACE) inhibitor; due to the fact that increased angiotensin in obese patients is thought to take a role in pathophysiology of ORG. Older two siblings with FSGS, managed to lose weight with dietary restriction and exercise; and their proteinuria is slightly reduced with therapy. But the youngest sibling aged 7.5, gained more weight and her proteinuria increased through the follow up.

Abstract GP72 Table 1

PATIENT	I	II	III
Age	7.5	12.5	16.5
Sex	F	F	F
Weight (kg)	35	84	92
BMI (percentile)	22 (97.7%)	34.5 (99%)	34.8 (98%)
Edema	(–)	(–)	(–)
Hypertension	(-)	(+)	(+)
24 hour proteinuria (mg/m2/h)	9.6	65	58
Creatinine (mg/dL)	0.35	0.44	0.89
Albumin (g/dL)	4.7	3.9	3.8
Uric asid (mg/dL)	3.7	6.7	9.8
eGFR	198	196	105
Urinary USG	Normal	Normal	Normal
Renal biopsy	Not diagnostic	FSGS	FSGS + chronic parenchymal injury
Weight change	+4 kg	−1 kg	−5 kg
Proteinuria after treatment (mg/m2/h)	17.1	31	44

Conclusion Obese children must be screened for proteinuria and ORG because early diagnosis, treatment with ACE inhibitors and weight loss can prevent the progression to end stage renal failure.

GP73 THE PHENOTYPE OF 8P23 DELETION SYNDROME

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Background 8p23 deletion syndrome is a unique chromosomal disorder that while rare, can have a variable spectrum of phenotypes. A pure 8p23 deletion is typically associated with microcephaly, developmental delay and congenital heart defects. It often occurs 'de novo' where part of the short p arm of chromosome 8 is missing. Some of these deletions are accompanied by a balanced translocation or rearrangement of chromosomes. There may be a gain/duplication of other genetic material. Our case is interesting in that we report a little girl who has an associated reciprocal duplication of 10q.

10q duplication syndrome is linked to a number of eye, kidney and craniofacial abnormalities depending on the how much extra chromosomal material there is.

Aim Our aim is to report a rare case of chromosomal 8p23 deletion with reciprocal 10q duplication due to a paternal balanced translocation in an 11 year old girl whose parents were originally given a diagnosis of Smith Lemi Opitz syndrome.

Methods We describe the clinical presentation, investigations and outcome to date of this patient in whom we confirmed a molecular diagnosis of the condition.

Results An 11 year old girl, born in India and given a diagnosis of Smith- Lemi-Opitz syndrome at birth, was referred to our Paediatric services by her GP, following her move to our region.

This complex little girl has significant global developmental delay, is visually impaired, non-verbal and non-ambulant. Her dysmorphic features include wave-shaped palpebral fissures, curly hair, long eyelashes, short philtrum, microcephaly, syndactyly and a repaired cleft palate.

She is the only child of non-consanguineous healthy parents.

Genetic studies including CGH array, have shown that she does not in fact have Smith-Lemi-Opitz syndrome but instead has an unusual 8p chromosomal deletion and 10q duplication.

Conclusion This case highlights the importance of early diagnosis and clinical follow-up of chromosomal disorders. This could lead to earlier intervention, access to specialists and as a result better outcomes in terms of the developmental potential of these children. Misdiagnosis we know, can often cause a delay in the appropriate treatment thus poorer quality of life in those affected. An accurate diagnosis will also allow the parents of this child to receive optimal genetic counselling should they decide to add to their family.

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