strong necessity to look for the precise familial history (sometimes hidden). -> Mullerian structures in AIS have already been reported. Some mechanisms for absence of Müllerian Ducts regression in AR mutation have been suggested as AMHR mutation., unavailable hypothethis in our patient. Further studies should promote a better understanding of this pathogenic association.

P2-d2-519 Sex Differentiation

The IVS1-2A>G mutation in the SRD5A2 gene is present in all Greek Cypriot patients with 5 alpha reductase deficiency diagnosed so far

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5 Alpha Reductase deficiency (5α SRD) is caused by mutations in the 5α Steroid Reductase type 2 gene (SRD5A2). More than 40 gene mutations have been reported worldwide, with some recurrent mutations commonly reported among various ethnic populations while other mutations reflect a founder gene effect in individuals with a common ancestry. The aim of our study was to identify the genetic defect in the SRD5A2 gene in Greek Cypriot patients with 5α SRD, whose diagnosis was based on clinical and biochemical criteria. Four unrelated patients with 46, XY karyotype and 5α SRD were investigated. Patients 1, 2 and 3 presented with ambiguous genitalia at birth whereas patient 4 (raised as girl) presented at the age of 14 years with signs of sexual infantilism and virilization. The hCG test was informative of 5α SRD, as it showed an elevated T/DHT ratio (23.5, 29 and 29.6) after stimulation. All five exons of the SRD5A2 gene were screened for mutations by direct sequencing of PCR products. Patients 1, 3 and 4 were found homozygous for the mutation A>G at the splice site of intron 1/exon 2 (IVS1-2A>G/IVS1-2A>G). Patient 2 was compound heterozygote for the mutations IVS1-2A>G and Pro181Leu. The same mutation (IVS1-2A>G) in the SRD5A2 gene was identified in four unrelated patients, both in homozygous and heterozygous state. This splice site mutation seems to characterize our population and is probably due to a gene founder effect. This underlying genetic abnormality has been already reported in Turkish patients and it may be characteristic for the Eastern Mediterranean region. The Pro181Leu mutation found most probably does not reflect a founder effect as it has been previously identified in patients of Italian origin.

P2-d2-520 Sex Differentiation

Three new mutations in androgen receptor gene identified in patients with 46,XY SDS: Complications for genetic counselling

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Androgen receptor insensitivity syndrome (AIS) is characterized by a wide spectrum of phenotypes ranging from 46,XY subjects with genitals completely female to normal male with infertility, more then 500 mutations are reported in the AR gene, mostly inherited in X-linked manner, few cases presented de novo mutations. In these last 20 years in vitro and in vivo functional studies on mutated androgen receptor, the determination of 3D structure of hormone binding domain, the identification of wide number of cofactors interacting with the androgen receptor, and the more recent studies of expression of genital tissue, gave a big contribute to understand the bio-molecular and pathogenetic aspects of androgen action. Here we reported the results of the analysis of AR gene in four patients with AIS, that we consider emblematic of how all these acquisitions on androgen receptor biology could help the clinical correlation of new mutations, but also highlight some critical aspects regarding in particular the genetic counselling to the families. All the patients has 46,XY karyotype, two are sisters with complete female genitals, diagnosed for the presence of inguinal hernia in the older sister, the other two are males with a severe hypospadias. The genomic DNA was isolated from peripheral blood and all coding sequences AR genes were submitted to direct sequencing. Four different mutations were found, three of which new, in one patient were identified two mutations. All the relatives available for the genetic analysis were studied for carrier identification, the results of families analysis revealed some incongruence regarding the mechanism of the mutagen event affecting the AR gene. On the basis of 3D structure and data reported in the literature we propose a genotype-phenotype correlation for the new mutations, with the purpose to give a contribute to clinical management and genetic counselling.

P2-d2-521 Sex Differentiation

Phalloplasty as a treatment for severe penile insufficiency

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Males with severe penile insufficiency in conditions such as penile agenesis, micropenis or some disorders of sex development (DSD) remain a major challenge to the reconstructive urologist. With a large experience using phalloplasty in female-to-male transsexual surgery, we started to use this technique for severe penile insufficiency. Eleven males (age 15 to 42 years) were treated with phalloplasty (7 with radial forearm free flap and 4 with anterolateral thigh flap) between March 2004 and December 2007 (follow-up: 3 to 47 months). All patients suffered psychologically from their condition with a low self-esteem and sexual and relational dysfunction. They were evaluated by a sexuologist-psychiatrist before and after surgery. Erectile implant surgery was offered about one year after the phallic reconstruction. There were no complications concerning the flap. Two complications (pulmonary embolism and severe hematuria) were reported in the early post-operative period. Four patients developed urinary complications (stricture and/or fistula). Patient satisfaction after surgery was high in 10 cases and moderate in one case. Psychological evaluation confirms this, especially on the self-esteem level. Six patients underwent erectile implant surgery. In 2 patients the erectile implant had to be removed due to infection or erosion. Our first experience with phalloplasty in young males has convinced us that this technique is a valuable treatment for severe penile insufficiency. It has good results on the self-esteem level and their sexual well-being. However, urinary complications occurred in 4/11 patients and explantation of the erectile implant occurred in 2/6 patients. Patients must be informed about these possible and frequent urological complications. This technique opens new horizons for the treatment of conditions such as penile agenesis, micropenis, some DSD conditions and cloacal exstrophy. More research is necessary on the criteria for patient selection, prevention of complications, and long-term functional and psychological outcome.

Patient	Indications	Type of phalloplasty	Age (years)	Follow-up (months)	Penile prosthesis
1	Shriveled penis - infected penile stiffener	Anterolateral thigh flap	42	38	AMS Ambicor, 2 cylinders
2	Shriveled penis - bladder extrophy	Radial forearm free flap	23	47	AMS Ambicor, 2 cylinders
3	Shriveled penis - bladder extrophy	Radial forearm free flap	16	40	AMS Ambicor, 2 cylinders
4	Penile amputation - epitheloid sarcoma	Radial forearm free flap	15	24	No
5	Crippled penis - hypospadias	Radial forearm free flap	20	24	No
6	Shriveled penis - bladder extrophy	Radial forearm free flap	15	22	No
7	Penile necrosis - traffic accident	Radial forearm free flap	32	37	AMS Ambicor, 2 cylinders
8	Cloacal extrophy	Anterolateral thigh flap	16	8	No
9	Cloacal extrophy	Anterolateral thigh flap	16	8	No
10	Micropenis - partial androgen insensitivity syndrome	Radial forearm free flap	30	13	No
11	Penile necrosis - priapism	Anterolateral thigh flap	38	3	No