

Bone mineral density in young women with complete androgen insensitivity syndrome. The AISIA study: preliminary data

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Background. A special issue in the management of individuals with complete androgen insensitivity syndrome (cAIS) is bone health. Bone mineral density (BMD) has been reported to be reduced, but data were collected in small number of patients or obtained by cumulating results from different machines, introducing difficulties in data interpretation. In addition, BMD data have been obtained in heterogeneous populations regarding gonadal status and/or substitutive therapy with the diagnosis based solely on clinical and/or endocrine features. In fact, the mutational analysis of androgen receptor has been not performed in several studies. Therefore, patients with phenotypes resembling AIS but affected by other disorders of sex development may be included. In addition, the efficacy of estrogen administration in normalizing BMD in women with cAIS and removed gonads remains unclear.

AIM. To evaluate BMD in women proven genetic diagnosis of cAIS.

Patients and methods. Nine young women (age 19-42 years) from AISIA (Italian support association for AIS) were enrolled in the study. Diagnosis of cAIS was made on the basis of clinical phenotype, 46,XY phenotype and mutational analysis of androgen receptor gene. Three patients were not gonadectomized at BMD scan, while the other ones were in post-pubertal period (age at gonadectomy 15.5 – 36.0 years). All the gonadectomized women were on hormonal substitutive therapy (HRT); compliance was graded as good (+), fair (+/-) or poor (-). BMD at lumbar spine and femoral neck were assessed by DXA (Lunar Prodigy). Values are expressed as T-score vs reference values.

Results. Better BMD, at both lumbar spine and femoral neck, was found in women without gonadectomy or with good compliance with HRT in comparison with those with fair or poor compliance (Figure).

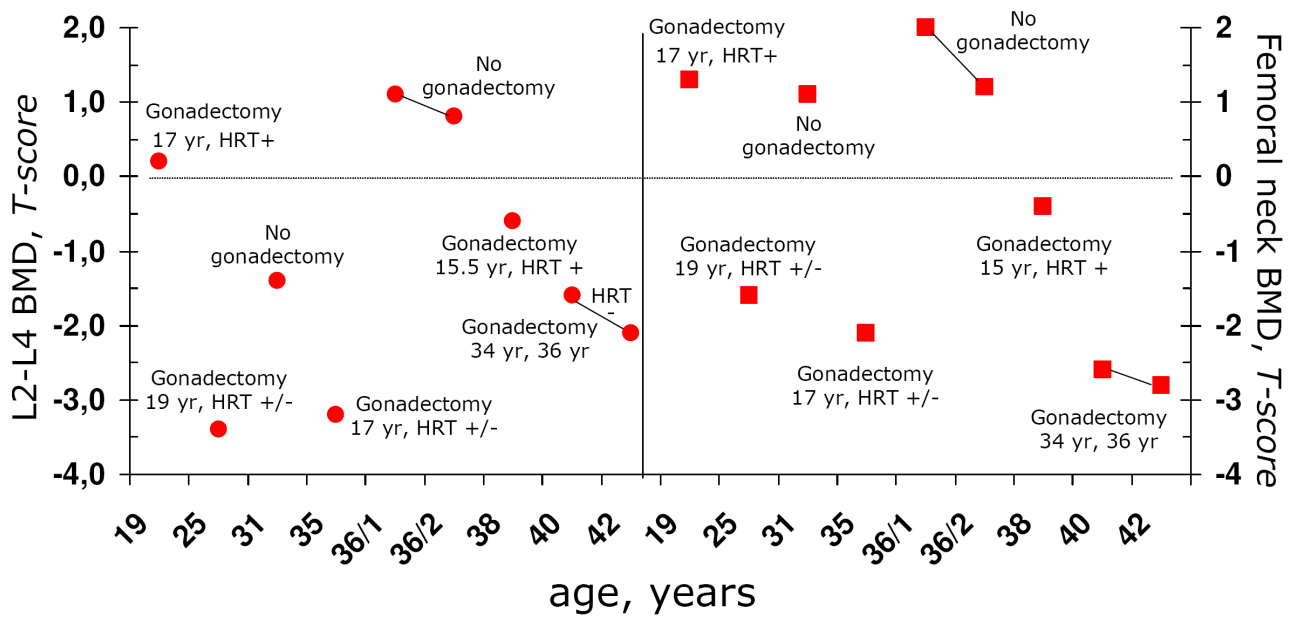


Figure. Areal BMD at lumbar spine (L2-L4) and at femoral neck in women with CAIS; data on gonadal status and compliance to HRT are shown for each individual.

Conclusion. Our preliminary data demonstrated that absence of gonadectomy or appropriate HRT and good compliance are associated with better bone status in young adult women with proven CAIS. Larger series of homogeneous patients will be investigated to obtain conclusive data and clear indications for the optimal management of people with CAIS.