

for clinical evaluation after an orthopedic surgeon reported placing an orthopedic pin with remarkable ease following a distal radial metaphyseal fracture sustained after a minor fall. She had previously sustained two fragility fractures involving the radius and ulna following a minor fall two years earlier. Her work up revealed none of the common characteristics of an adolescent female with osteoporosis or a fragility fracture. Her previous x-rays were reviewed and demonstrated an unusual finding. Treatment outcome is discussed.

Results: Her clinical evaluation revealed no evidence of genetic, endocrine, neoplastic, dietary or psychiatric disease. She had never utilized anticonvulsants, heparin, or corticosteroids, nor were there any significant identifiable environmental exposures. Serum and urinary makers of bone formation and bone resorption were elevated. Dual energy x-ray absorptiometry (DEXA) demonstrated severe osteopenia of the hip, spine, and radius (Z-scores ranging from -1.75 to -2.0). Skin biopsy revealed no evidence of a type I collagenopathy, effectively ruling out osteogenesis imperfecta. Review of x-rays performed 2 months after the most recent fracture revealed "neo-osseous osteoporosis", the complete healing of the fracture with notable absence of increased mineralization and callus formation. This has been described as the most characteristic finding in adolescents with IJO. After treatment with risedronate, calcium, and vitamin D, a marked decrease in serum osteocalcin and urinary deoxypyridinoline was noted. Significant improvement in bone density was demonstrated by repeat DEXA scanning one year later. No new fragility fractures were sustained in this interval.

Conclusions: IJO is a rare disease of adolescence and childhood characterized by osteoporosis with total or near total spontaneous remission after puberty. Fewer than 100 cases have been described in the world literature. The etiology is unknown and the diagnosis of IJO is largely one of exclusion of other etiologies. Risedronate was well tolerated in this 14-year-old female and the substantial decrease in bone turnover markers in the first 3 months of treatment suggests an important role for bisphosphonates in the treatment of adolescent osteoporosis. Randomized controlled trials are indicated to further define the role of bisphosphonates in the treatment of adolescent osteoporosis. The effect of bisphosphonates on future reproductive outcomes also needs to be established. Gynecologists should be aware of the presence and various etiologies of osteoporosis in the adolescent female.

Tuesday, October 23, 2001
3:00 P.M.

O-146

Secondary amenorrhea in a 46,XY female. S. E. Pollack, R. A. Bennett. Albert Einstein Coll of Medicine, Bronx, NY.

Objective: To describe a case report of the presentation, workup and treatment of a secondary amenorrhea patient found to be genotypically 46,XY.

Design: A case report.

Materials/Methods: Review of the Medline literature from 1966–2001.

Results: A 27-year-old nulligravid, sexually active, Dominican female presented to our office complaining of infertility and amenorrhea of one year's duration. The patient stated that menarche occurred at age 13, with subsequent menses every 30 to 90 days. She did complain of premenstrual cramping and mastalgia. Physical examination revealed her to be 66 inches and 170 pounds. Tanner staging was stage 3 breasts and stage 5 pubic hair. She did have some mild Turner syndrome stigmata, including widely spaced nipples and cubitus valgus. Pelvic examination revealed normal female external genitalia, a normal vagina, a small cervix and uterus, and nonpalpable gonads. Pelvic ultrasound and magnetic resonance imaging revealed a premenarcheal uterus and nonvisualized gonads. Laboratory studies, including androgens, came back in normal reproductive female ranges, except for a follicle stimulating hormone level of 71 mIU/ml and an estradiol of 17 pg/ml. A peripheral blood karyotype demonstrated a 46,XY genotype. Y chromosome probes revealed a completely intact Y chromosome. The patient was then taken to the operating room where a laparoscopic gonadal extirpation was performed. Pathology was consistent with bilateral streak ovaries comprised of scant ovarian stroma devoid of follicles. Post operatively, bone densitometry revealed osteoporosis, and the patient was put on oral contraceptives as hormone replacement. She was counseled regarding oocyte donation as a treatment for her infertility.

Conclusions: To our knowledge, ours is the only patient in the medical

literature who is a 46,XY female presenting with secondary amenorrhea who has a proven intact Y chromosome. There are only four other reported cases of 46,XY females with secondary amenorrhea. Study of this patient has the potential to help define as yet unknown mechanisms of sex determination.

REPRODUCTIVE BIOLOGY

Tuesday, October 23, 2001
2:00 P.M.

O-147

Expression of telomerase activity in mouse oocytes and embryos. T. Kaneko, H. Saito, S. Kawachiya, T. Saito, H. Kurachi. Yamagata Univ Sch of Medicine, Yamagata City, Japan.

Objective: Telomeres are the distal ends of chromosomes composed of tandem repeats of the sequence TTAGGG. Possible functions of telomeres include prevention of chromosome degradation, end-to-end fusions, rearrangements, and chromosome loss. Telomerase is a ribonucleoprotein that synthesizes telomeric DNA for addition to the end of the chromosome using a segment of its RNA component as a template. Telomerase, counteracts the mitotic clock of telomere shortening. It has been hypothesized that telomerase activity is necessary for cellular immortalization and that telomerase activity is present in cells of germline origin. The objective of the present study was to determine the level of telomerase activity in oocytes and individual blastomere of early embryos, and also to assess the correlation between the telomerase activity and embryo development potential.

Design: Telomerase activity was determined in oocytes and embryos from B6C3F1 mice by modified TRAP assay kit.

Materials/Methods: Five to six week old female B6C3F1 mice were used for the experiment. Oocytes were recovered following a conventional PMSG/hCG superovulation protocol. Oocytes were recovered by aspirating follicles at 24 h (P24), 48 h (P48) after PMSG and at 4 h (H4), 8h (H8) after hCG. Oocytes and embryos were collected from the oviducts at 14 h (H14), 2 days (2 cell), 3 days (morula), 4 days (blastocyst) after hCG and mating. Immature intrafollicular oocytes were denuded of cumulus cells by hyaluronidase treatment. Collected oocytes and embryos were immediately snap-frozen and stored at -80°C until assayed. For the measurement of the telomerase activity, TeloChaser of TOYOBO Co. (Telomerase assay kit by Stretch PCR method) was used.

Results: Telomerase activity was found in all stages of the intrafollicular oocytes and ovulated oocytes. In the unfertilized oocyte, the telomerase activity of P24 was high, and it was lower at P48, H4 and H8. It started to increase again at H14. In the fertilized oocyte, the activity weakened from 1 cell to morula, and increase strongly back at blastocyst stage.

Conclusions: These data demonstrate that telomerase activity is present in germ cells at various stages of development, and suggest that telomerase activity may be important for meiosis and mitosis. The high levels of telomerase activity in the intrafollicular oocytes and blastocyst may serve as a marker for monitoring the effects of hormonal agents, aging, and toxins on oocyte and embryo quality.

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Tuesday, October 23, 2001
2:15 P.M.

O-148

Inheritance of donor versus recipient mitochondrial populations in human offspring derived from ooplasmic transplantation. K. E. Pierce, J. A. Barritt, J. Cohen, C. A. Brenner. Brandeis Univ, Waltham, MA; Institute for Reproductive Medicine and Science of Saint Barnabas, West Orange, MA; Institute for Reproductive Medicine and Science of Saint Barnabas, West Orange, NJ.

Objective: Mitochondrial DNA heteroplasmy, a mixture of donor and patient mitochondrial DNA (mtDNA) has been detected in placenta, fetal cord blood and infant bloods in only two of twelve babies assessed after cytoplasmic transfer. Mitochondrial DNA fingerprinting of the hypervariable region suggests that the percentage of donor mitochondria might vary