



AN OPT-OUT APPROACH TO EXPANDED CARRIER SCREENING (ECS) INFORMS PATIENT AND CLINICIAN DECISION MAKING AND IDENTIFIES MORE PREIMPLANTATION GENETIC DIAGNOSIS (PGD) APPROPRIATE CASES

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OBJECTIVE: Patients seek fertility care to increase the probability of having a healthy child. ACOG recently published committee opinions which highlight the use of ECS in the era of genomic medicine as a crucial part of preconception counseling that should be offered before a pregnancy is conceived. For patients seeking infertility care a unique opportunity exists for this counseling, screening, and, if appropriate, treatment in the form of PGD. We sought to determine whether an opt-in or an opt-out approach to ECS identified more carriers and PGD appropriate cases.

DESIGN: Retrospective cohort study

MATERIALS AND METHODS: All new infertility patients seen at a large academic affiliated private practice from quarter 1 of 2011 to quarter 4 of 2016 were included. Until the 3rd quarter of 2015 the practice employed an opt-in approach to ECS and offered ECS as an option in addition to standard ethnic-based preconception testing. The ECS test utilized was dictated by patient preference and/or insurance coverage. During the 3rd quarter of 2015 the practice changed policy to an opt-out approach to ECS and patients were required to decline that method of testing. The default test was a sequencing-based ECS along with follow-up discussion with a genetic counselor.

RESULTS: Over the study period 24,329 new infertility patients were seen and 18,541 ECS tests were ordered, including tests for both the male and female partner. Of these, 2,449 tests were not performed, typically because the patients decided not to pursue further treatment. The rate of incomplete tests was higher once the opt-out policy was implemented ($p < 0.05$). There were 15,880 ECS tests with results, 5,738 of which were positive for one or more condition. There was an increase in the rate of positive test per ECS ordered after the opt-out policy was initiated with use of the sequencing-based ECS as the default. Finally, of the couples tested, there was an increased rate of PGD recommendation as part of the first IVF cycle plan after implementation of the opt-out approach ($p < 0.05$).

CONCLUSIONS: These data suggest that an opt-out approach to ECS allows physicians and patients to have a more comprehensive and informed preconception work-up and results in the opportunity for couples to have the option for PGD in an increased number of cases. With nearly 15,000 documented genes involved in human disease, genomic medicine represents an area of rapid growth and innovation. ECS allows fertility treatment teams to be involved on the front line of this innovation and empower patients in their decisions to build a healthy family. An opt-out approach to ECS as a part of preconception counseling presents a method of balancing the burden of care with the benefit provided through increased knowledge and ability for intervention with PGD when appropriate.