Ethical considerations of 'mainstreaming' genetic testing services to non-geneticist physicians: a focus on counselling and informed consent

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Background and Objectives: The growth in genetic technology and increasing applications of genetics-driven practices across all areas of medicine have led to increasing demands for genetic testing services. These demands and the relative scarcity of medical geneticists available to meet these demands have resulted in a movement towards "mainstreaming" genetic testing services, which is when genetic testing services are performed by non-geneticist clinicians. There have been several concerns about the ethics of non-geneticist clinicians delivering these services. The present project sought to expand upon previously identified ethical considerations of mainstreaming by critically reviewing the literature, discussing the work that exists currently and identifying gaps where more research and ethical reflection are needed. Specifically, this project focuses on the ethical considerations of mainstreaming the counselling and informed consent of genetic testing services.

Methods: A critical review was performed to evaluate the ethical considerations of mainstreaming genetic testing services to non-geneticist physicians. The initial search was performed in Medline, Scopus and Embase, and reference list searching helped to further identify articles for inclusion. Articles were selected for inclusion based on how they contribute to the research question: What are the ethical considerations of mainstreaming the counselling and informed consent of genetic testing services to non-geneticist physicians?

Results: Selected articles and the ethical considerations therein were analyzed and discussed, drawing on principles of biomedical ethics. Topics of discussion explored include: differences in informed consent and counselling practices between geneticist and non-geneticist physicians; the need for specific guidelines that inform mainstream counselling and informed consent for genetic testing in a specific setting, specialty or in the context of a specific medical condition; the changing role of the non-geneticist physician and the limits of the non-geneticist's role; guidance for documenting counselling and consent discussions; and the need for more work to ensure equity in the provision of counselling and informed consent for mainstream genetic testing.

Conclusions: As mainstreaming continues to be implemented across different areas of medicine, it will be crucial to identify and explore ethical considerations to reduce harm that could arise as a result of transitioning to this practice.

Keywords: mainstreaming; genetics; ethics

The Effects of 20-Hydroxyecdysone (Ecdysterone) on Athletic and Physiologic Outcomes: A Systematic Review of Randomized Controlled Trials

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Background and Objectives: 20-Hydroxyecdysone (20E) is a phytoecdysteroid with emerging use as an anabolic agent amongst athletes. A systematic review was performed for the purpose of evaluating the clinical and athletic outcomes of 20E administration (in the form of purified and semipurified extracts) on physical performance, body composition, endocrine/metabolic and safety outcomes in adult populations.

Methods: The review was guided by the Cochrane Handbook for Systematic Reviews (of interventions). Randomized and controlled studies in adults receiving oral 20E or 20E rich extracts with or without structured training were included. All in vivo/in vitro studies, narrative reviews and non interventional studies were excluded. Literature search was conducted with database searches and manual review. A narrative synthesis was conducted and organized by domain of outcome.

Results: The risk of bias of selected studies ranged from low to moderate. Across athletic young males, older adults with sarcopenia, competitive athletes, and overweight individuals, 20E doses from 3 to 350 mg twice daily over 10 days to 9 months generally improved at least one performance, body composition, or metabolic parameter versus placebo or control. Outcomes included significant improvements in strength, gait speed, preservation of muscle mass during detraining, visceral fat reduction, increased fat oxidation, favorable changes in cortisol, insulin sensitivity, and cardiorespiratory function. Safety outcomes demonstrated no systemic toxicity or endocrine disruption, with a phase 1 showing favorable safety outcomes of doses as high as 1400 mg 20E per day.

Discussion: The current human evidence supports a role of 20E in improving exercise and body composition outcomes in adults, with a consistent signal being demonstrated as improvements of strength / power, gait speed/functional mobility and body composition.

Some trials noted favorable changes in metabolic and endocrine markers, others noting strong signals for improved cardiopulmonary function in comorbid populations (obesity, COVID-19).

A significant limitation in this review was the differing study populations and designs, small sample sizes, and lack of long term data. The certainty of efficacy is low to moderate

Conclusions: The evidence supports a benefit of 20E supplementation for athletic applications and improved exercise outcomes in non athletic adult populations. Larger randomized control trials with similar dosing/endpoints in strength and body composition are needed to quantify the effect size of 20E supplementation on athletic outcomes.

Keywords: 20-hydroxyecdysone; physical performance; systematic review