AMERICAN MEDICAL ASSOCIATION HOUSE OF DELEGATES

Resolution: 507
(A-23)

Introduced by: Medical Student Section

Subject: Recognizing the Burden of Rare Disease

Referred to: Reference Committee E

Whereas, Rare diseases, also known as orphan diseases, are defined as conditions that affect less than 200,000 individuals in the United States (US), categorized into various overlapping disease classes including but not limited to chromosomal disorders, connective tissue diseases, blood diseases, metabolic disorders, skin diseases, and autoimmune conditions[1-3]; and

Whereas, Rare diseases cumulatively affect a significant number of people in the US, estimated to be between 25-30 million individuals[4]; and

Whereas, Congress passed The Orphan Drug Act (ODA) to incentivize drug companies to develop treatments for rare diseases and rapidly deploy novel agents to target conditions affecting fewer than 200,000 persons in the United States, or conditions for which a drug will not be profitable within 7 years following approval by the FDA[5-7]; and

Whereas, Current orphan drug legislation to support biopharmaceutical R&D portfolio diversity, enhance patent exclusivity, and provide distinct FDA designations is not sufficient to promote novel drug development for different rare disease classes as 90% of these patients are without an FDA approved treatment[8-10]; and

Whereas, The Affordable Care Act does not specifically address orphan drugs coverage, and even when new treatment options such as drug prescriptions or medical devices are available for people with rare diseases, 61% of patients are denied or delayed in accessing treatment due to insurance company pre-approval[8,11]; and

Whereas, There are many disparities in rare disease health care including 39% of respondents traveling 60 or more miles for medical care, 17% considering or completing relocation, and 29% being granted access to treatment not approved by FDA[1,8]; and

Whereas, The mean health related quality of life scores of those with orphan diseases were the poorest compared to individuals with common chronic diseases, which may be attributed to diagnostic challenges, decreased access to medical information and treatment, and negative psychological impact such as coping with uncertainty[12]; and

Whereas, In 2019, health care costs associated with orphan diseases may be comparable to heart disease or cancer at $966 billion, accounting for direct, indirect, and non-medical costs associated with diagnosis and amounting to nearly 50% of the total national bill, despite a vastly lower percentage of rare disease within the population[10,13,14]; and

Whereas, The number of documented cases of many rare diseases are only expected to increase given recent advances in genomics and personalized medicine[15]; and

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Whereas, There is a lack of reliable epidemiological data for patients with orphan diseases and insufficient knowledge on the pathophysiology of these conditions among health care providers, leading to inadequate access to information on disease prevalence and treatment outcomes[16]; and

Whereas, A lack of knowledge has made treatment options difficult for patients with orphan diseases to access, contributing to difficulty and delay in diagnosis, as shown by a National Organization for Rare Diseases (NORD) 2019 report that found 28% of individuals diagnosed with a rare disease did not receive a diagnosis for seven years or more and 38% of individuals received a misdiagnosis[8,17,18]; and

Whereas, Due to barriers in accessing treatment options, patients with rare diseases have difficulty finding treatment information and patient registries, such as Rare Disease Registries have become a tool for both patients and physicians to be educated on their condition[19]; and

Whereas, Natural history studies and patient registries collecting longitudinal, patient-driven data aided by machine learning help advance our understanding of rare diseases and how they progress over time, facilitating clinical research and the development of novel therapeutics[8,20]; and

Whereas, Recent automated tracking systems, such as RENEW, are being used to gather new global genomic discoveries onto an accessible database for genome sequencing of patients for improved therapeutic outcomes[21]; and

Whereas, Incorporation of genomic research as clinical diagnostic tests can increase large scale sequencing projects of structural variants and sharing of data that shortens the time to diagnosis by producing increased cohort sizes for development of personalized therapeutic options[22]; and

Whereas, With future advances in techniques such as genome-wide pooled CRISPR screening and plasmid-based reporter assays, which can shorten time to diagnosis, precision therapeutics could be used as a targeted and efficient approach in orphan disease treatment[23,24]; and

Whereas, With only 30% of the genome accounted for in the diagnosis of rare disease there is still 75% of phenotypic variations within the genome unaccounted for, in which future novel gene discovery through sequencing efforts can overcome this diagnostic challenge[24]; and

Whereas, AMA policy H-185.963 emphasizes insurance coverage for childhood and congenital diseases, but does not sufficiently include the orphan disease population or specialized genomic research considerations needed for timely diagnosis and treatment; therefore be it

RESOLVED, That our American Medical Association recognize the under-treatment and under-diagnosis of orphan diseases, the burden of costs to health care systems and affected individuals, and the health disparities among patients with orphan diseases (New HOD Policy); and be it further

RESOLVED, That our AMA support efforts to increase awareness of patient registries, to improve diagnostic and genetic tests, and to incentivize drug companies to develop novel therapeutics to better understand and treat orphan diseases. (New HOD Policy)

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Fiscal Note: Minimal - less than $1,000

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RELEVANT AMA POLICY

Genetic Information and Insurance Coverage H-185.972
AMA believes: (1) Health insurance providers should be prohibited from using genetic information, or an individual's request for genetic services, to deny or limit any health benefit coverage or establish eligibility, continuation, enrollment or contribution requirements.
(2) Health insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information or an individual's request for genetic services.
(3) Health insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information.
(4) Health insurance providers and other holders of genetic information should be prohibited from releasing genetic information without express prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure would be made.
Citation: BOT Rep. 15, I-96; Reaffirmed: CMS Rep. 8, A-06; Reaffirmed in lieu of Res. 102, A-10; Reaffirmation: A-17; Reaffirmed: BOT Rep. 12, I-21;

Insurance Coverage for Adults with Childhood Diseases H-185.963
Our AMA: (1) urges public and private third party payers to increase access to health insurance products for adults with congenital and/or childhood diseases that are designed for the unique needs of this population; and
(2) emphasizes that any health insurance product designed for adults with congenital and/or childhood diseases include the availability of specialized treatment options, medical services, medical equipment and pharmaceuticals, as well as the accessibility of an adequate number of physicians specializing in the care of this unique population.
Citation: CMS Rep. 2, I-99; Modified and Reaffirmed: CMS Rep. 5, A-09; Reaffirmed: CMS Rep. 01, A-19;

Coverage of Children's Deformities, Disfigurement and Congenital Defects H-185.967
1. The AMA declares: (a) that treatment of a minor child's congenital or developmental deformity or disorder due to trauma or malignant disease should be covered by all insurers; (b) that such coverage shall include treatment which, in the opinion of the treating physician, is medically necessary to return the patient to a more normal appearance (even if the procedure does not materially affect the function of the body part being treated); and (c) that such insurability should be portable, i.e., not denied as a pre-existing condition if the patient's insurance coverage changes before treatment has been either initiated or completed.
2. Our AMA will advocate for appropriate funding for comprehensive dental coverage (including dental implants) for children with orofacial clefting.
Citation: (Sub. Res. 119, I-97; Reaffirmed, A-03; Reaffirmation A-05; Reaffirmation A-08; Appended: Res. 109, A-13)

Addressing Financial Incentives to Shop for Lower-Cost Health Care H-185.920
1. Our AMA supports the following continuity of care principles for any financial incentive program (FIP):
a. Collaborate with the physician community in the development and implementation of patient incentives.
b. Collaborate with the physician community to identify high-value referral options based on both quality and cost of care.
c. Provide treating physicians with access to patients' FIP benefits information in real-time during patient consultations, allowing patients and physicians to work together to select appropriate referral options.
d. Inform referring and/or primary care physicians when their patients have selected an FIP service prior to the provision of that service.
e. Provide referring and/or primary care physicians with the full record of the service encounter.
f. Never interfere with a patient-physician relationship (eg, by proactively suggesting health care items or services that may or may not become part of a future care plan).
g. Inform patients that only treating physicians can determine whether a lower-cost care option is medically appropriate in their case and encourage patients to consult with their physicians prior to making changes to established care plans.
2. Our AMA supports the following quality and cost principles for any FIP:
a. Remind patients that they can receive care from the physician or facility of their choice consistent with their health plan benefits.
b. Provide publicly available information regarding the metrics used to identify, and quality scores

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associated with, lower and higher-cost health care items, services, physicians and facilities.
c. Provide patients and physicians with the quality scores associated with both lower and higher-cost physicians and facilities, as well as information regarding the methods used to determine quality scores. Differences in cost due to specialty or sub-specialty focus should be explicitly stated and clearly explained if data is made public.
d. Respond within a reasonable timeframe to inquiries of whether the physician is among the preferred lower-cost physicians; the physician's quality scores and those of lower-cost physicians; and directions for how to appeal exclusion from lists of preferred lower-cost physicians.
e. Provide a process through which patients and physicians can report unsatisfactory care experiences when referred to lower-cost physicians or facilities. The reporting process should be easily accessible by patients and physicians participating in the program.
f. Provide meaningful transparency of prices and vendors.
g. Inform patients of the health plan cost-sharing and any financial incentives associated with receiving care from FIP-preferred, other in-network, and out-of-network physicians and facilities.
h. Inform patients that pursuing lower-cost and/or incentivized care, including FIP incentives, may require them to undertake some burden, such as traveling to a lower-cost site of service or complying with a more complex dosing regimen for lower-cost prescription drugs.
i. Methods of cost attribution to a physician or facility must be transparent, and the assumptions underlying cost attributions must be publicly available if cost is a factor used to stratify physicians or facilities.
3. Our AMA supports requiring health insurers to indemnify patients for any additional medical expenses resulting from needed services following inadequate FIP-recommended services.
4. Our AMA opposes FIPs that effectively limit patient choice by making alternatives other than the FIP-preferred choice so expensive, onerous and inconvenient that patients effectively must choose the FIP choice.
5. Our AMA encourages state medical associations and national medical specialty societies to apply these principles in seeking opportunities to collaborate in the design and implementation of FIPs, with the goal of empowering physicians and patients to make high-value referral choices.
6. Our AMA encourages objective studies of the impact of FIPs that include data collection on dimensions such as:
a. Patient outcomes/the quality of care provided with shopped services;
b. Patient utilization of shopped services;
c. Patient satisfaction with care for shopped services;
d. Patient choice of health care provider;
e. Impact on physician administrative burden; and
f. Overall/systemic impact on health care costs and care fragmentation.
Citation: CMS Rep. 2, I-19;

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