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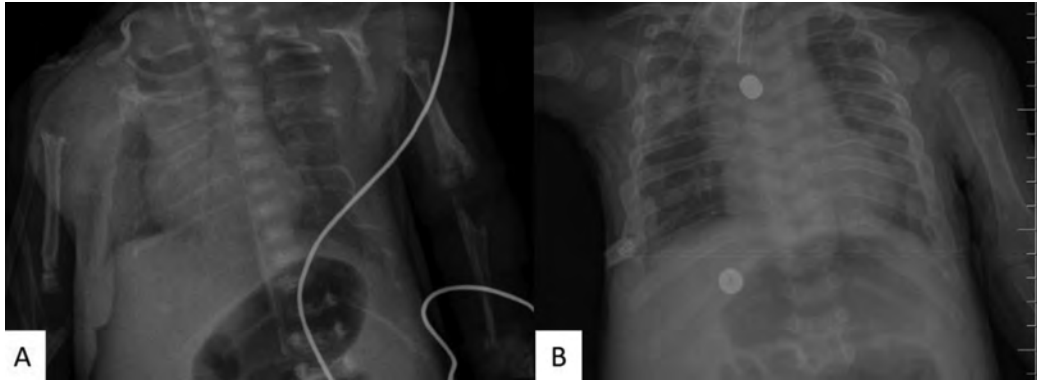
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General improvement before and 12 months after treatment with asfotase alfa

A Case of the Perinatal Form Hypophosphatasia Caused by a Novel Large Duplication of the *ALPL* Gene and Report of One Year Follow-up with Enzyme Replacement Therapy

Hacıhamdioğlu B et al.

DOI: 10.4274/jcrpe.galenos.2018.2018.0217



Official Journal of
Turkish Pediatric Endocrinology
and Diabetes Society

GoQuick™

Genotropin (rekombinant somatropin)
Kullanıma hazır kalem

kolay¹

güvenli¹

kullanıma hazır^{2,3}

5.3mg
12mg^{2,3}

Referanslar: 1. Hey-Hadavi J et al. Clin Ther. 2010;32:2036-47. 2. Genotropin® GoQuick™ 16 IU (5.3 mg) Kısa Ürün Bilgisi. 3. Genotropin® GoQuick™ 36 IU (12 mg) Kısa Ürün Bilgisi.

Genotropin® Kısa Ürün Bilgisi Özeti: GENOTROPIN GOQUICK® 16 IU (5.3 mg) - 36 IU (12 mg) enjeksiyonluk solüsyon için toz ve çözücü iğneri kullanıma hazır kalem. **Formül:** Rekombinant DNA teknolojisiyle Escherichia Coli hücrelerinde üretilmiş 16 IU (5.3 mg) - 36 IU (12 mg) somatropin içerir. **Endikasyonları:** Büyüme hormonunun yetersiz salgılanmasına bağlı çocuklardaki büyüme bozukluklarında; gonadal disgenesi (Turner Sendromu) ile birlikte bulunan büyüme bozukluklarında; kronik böbrek yetersizliği olan prepubertal çocuklardaki büyüme bozukluklarında; SGA tedavisinde - doğum ağırlığı ve/veya uzunluğu -2 SD olan ve 4 yaş ve sonrasında gerekli büyüme yakalayamamış (son 1 yılda yıllık boy kazanımı SDS<-0) çocuklarda veya gestasyonel yaşına göre küçük doğmuş olan (SGA) kısa çocuklardaki büyüme bozukluklarında (uzunluk SDS<-2.5 ve ebeveyn uyarlanış uzunluk SDS<-1) - ; hipotalamus-hipofiz hastalığı saptanan hipofizer cerrahi girişim geçirmiş, kranial radyoterapi görmüş veya çocuklukta başlamış büyüme hormonu yetmezliği olan erişkinler ile hipofizde adenomu olan hastalarda büyüme hormonu eksikliği varsa veya büyüme hormonu yetersizliğini düşündürülen bulguların bulunması durumunda biyokimyasal tanı testleri ile büyüme hormonu eksikliği kesin olarak saptanan yetişkinlerde, özellikle: konjenital veya idiyopatik hipofiz hastalıkları, hipotalamus-hipofiz tümörleri ve tedavileri sonrası, kraniofarengioma tedavisinden sonra, cerrahi girişim hasarlarında, Sheehan sendromu ve vasküler sebeple gelişen iskemik sebebli büyüme hormonu yetersizlikleri, radyasyon, travma, kronik otoimmün, bakteriyel veya viral enflamasyonlar ile hemokromatozis ve amiloidozisde görülen hipofizer yetmezliklerde, septo-optik displaziye meydana gelebilen aşikâr büyüme hormonu eksikliğinin replasmanı için büyüme hormonu tedavisi endikasyonu vardır. **Pozoloji:** Çocuklardaki büyüme hormonu salgılanma yetersizliğine bağlı büyüme bozukluğunda: Genellikle 0.025 - 0.035 mg/kg veya 0.7 - 1.0 mg/m² önerilmektedir. Turner Sendromuna bağlı büyüme bozukluğu: 0.045-0.050 mg/kg veya 1.4 mg/m² önerilir. Kronik böbrek yetmezliğine bağlı büyüme bozukluğu: 0.045-0.050 mg/kg (1.4 mg/m²) önerilir. Büyüme hızı çok düşüğe daha yüksek dozlar gerekebilir. Gestasyonel yaşa göre küçük doğmuş (SGA) olan kısa boylu çocukların büyüme bozukluklarında: Final uzunluğa erişinceye kadar genellikle vücut ağırlığına göre günlük 0.035 mg/kg (1.0 mg/m²) önerilmektedir. Yetişkinlerdeki büyüme hormonu eksikliği: Çocukluk çağı BHY sonrasında büyüme hormonu tedavisine devam eden hastalarda önerilen yeniden başlangıç dozu 0.2-0.5 mg/gün' dür. Yetişkin başlangıçlı BHY olan hastalarda tedavi düşük doz (0.15-0.3 mg/gün) ile başlanmalıdır. **Uygulama şekli:** Enjeksiyonlar subkutan enjeksiyon şeklinde ve lipotrofi gelişmesini önleyebilmek için her seferinde yeri değiştirilerek uygulanır. **Kontrendikasyonlar:** Elkin madde veya yardımcı maddelerden herhangi birine karşı aşırı duyarlılık durumunda kullanılmamalıdır. Somatropin, tümör aktivitesini gösteren herhangi bir bulgunun bulunması durumunda kullanılmamalıdır. Büyüme hormonu tedavisine başlanmadan önce intrakranial tümörler inaktif olmalı ve antitümör tedavi tamamlanmış olmalıdır. Tümör büyümesine ilişkin kanıt olması halinde tedavi sonlandırılmalıdır. GENOTROPIN GOQUICK® epifizleri kapanmış çocuklarda büyümenin uyarılması için kullanılmamalıdır. Açık kalp ameliyatı, abdominal cerrahi, kazaya bağlı multipl travma, akut solunum yetmezliği veya benzeri durumları izleyen komplikasyonların bulunduğu akut kritik hastalığı olan hastalara GENOTROPIN GOQUICK® uygulanmamalıdır. **Özel kullanım uyarıları ve önlemleri:** Hastalığın tanısı ve GENOTROPIN GOQUICK® tedavisi, terapötik kullanım endikasyonunda; hastaların tanı ve tedavisinde yeterli nitelikte ve tecrübeli doktorlar tarafından başlatılmalı ve takip edilmelidir. Maksimum önerilen günlük doz aşılmalıdır. Miyozis çok nadir bir advers olaydır ve koruyucu madde metakrezol ile ilişkili olabilir. Somatropin insülin hassasiyetini azaltabilir. Diabetes mellitus olan hastalarda somatropin tedavisine başlandıktan sonra insülin dozunun ayarlanması gerekebilir. Büyüme hormonu T4'ün T3'e tiroit dışı dönüşümünü artırabilir ve bu durum serum T4'ünün azalmasına ve serum T3'ünün artmasına yol açabilir. Malign bir hastalığın tedavisine sekonder büyüme hormonu yetersizliğinde malignitenin relaps belirtilerine dikkat edilmesi önerilmektedir. Çocukluk döneminde kanser sonrası sağkalımlarda, somatropin ile tedavi edilen hastalarda ilk neoplazma sonrası ikinci bir neoplazma gelişiminde risk artışı bildirilmiştir. Büyüme hormonu yetersizliği dahil, endokrin bozukluğu olan hastalarda kalça ekleminde epifiz kayması genel popülasyondan daha sık görülebilir. Sıddetli veya tekrarlayan baş ağrısı, görme sorunları, bulantı ve/veya kusma gelişmesi halinde papilla ödemi için funduskopi yapılması önerilmektedir. Somatropin iğneri her seferinde kullanıma hazır olarak sunulmaktadır. İlaç Etkileşimleri: Glukokortikoidlerle eş zamanlı tedavi somatropin iğneri ürünlerin büyüme hormonu etkileşimlerini engelleyebilir. Büyüme hormonu eksikliği olan yetişkinlerde yapılan bir etki çalışmasında somatropin uygulamasının sitokrom P450 1A2 izoenzimiyle metabolize olduğu bilinen bileşimlerin klirensini artırdığı belirtilmiştir. **Gebelik kategorisi:** C. **İstenmeyen etkiler:** Enjeksiyon bölgesi reaksiyonları, artralji, periferik ödem, parastezi, karpal tünel sendromu, miyalji, kas-iskelet sertliği çok yaygın ve yaygın görülen istenmeyen etkilerdir. **Doz aşımı ve tedavisi:** Akut doz aşımı başlangıçta hipoglisemi ve takiben hiperglisemiye neden olabilir. Uzun süreli doz aşımı fazla miktarda insan büyüme hormonunun bilinen etkilerine benzer belirtiler ve bulgulara neden olabilir. **Saklama koşulları:** Soğutulmuş (2°C - 8°C'de) saklayınız. Dondurmayınız. İki kompartımanlı kartuşlu/önceden doldurulmuş kalemli iğneri saklamak için dış kutusunda saklayınız. **Ticari Takdim Şekli ve Ambalaj Muhtevası:** 16 IU, 36 IU GoQuick® enjeksiyonluk solüsyon için toz ve çözücü iğneri 1 adet kullanıma hazır kalem. Reçete ile satılır. **Satış Fiyatı:** GoQuick® 16 IU 273.84 TL (19.02.2019), GoQuick® 36 IU 617.43 TL (19.02.2019). Sosyal Güvenlik Kurumu tarafından geri ödenir. Ödeme koşulları ile ilgili detaylı bilgi için Sosyal Güvenlik Kurumu, Sağlık Uygulama Talimatına bakınız. **Kısa ürün bilgisi/ kullanma talimatı onay tarihi:** 03.11.2016 **Ruhsat No:** 103/41 **Ruhsat tarihi:** 18.12.1997 **Ruhsat sahibi:** Pfizer İlaçları Ltd. Şti. Muallim Naci Cad. No:55 34347 Ortaköy-İSTANBUL Telefon no: (212) 310 70 00 - Faks no: (212) 310 70 58. Daha geniş bilgi için firmamıza başvurunuz: www.pfizer.com.tr

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All manuscripts must adhere to the limitations, as described below, for text only; the word count does not include the abstract, references, or figure/table legends. The word count must be noted on the title page, along with the number of figures and tables. Original Articles should be no longer than 5000 words and include no more than six figures and tables and 50 references.

Short Communications are short descriptions of focused studies with important, but very straightforward results. These manuscripts should be no longer than 2000 words, and include no more than two figures and tables and 20 references.

Brief Reports are discrete, highly significant findings reported in a shorter format. The abstract of the article should not exceed 150 words and the text/article length should not exceed 1200 words. References should be limited to 12, a maximum of 2 figures or tables.

Clinical Reviews address important topics in the field of pediatric endocrinology. Authors considering the submission of uninvited reviews should contact the editors in advance to determine if the topic that they propose is of current potential interest to the Journal. Reviews will be considered for publication only if they are written by authors who have at least three published manuscripts in the international peer reviewed journals and these studies should be cited in the review. Otherwise only invited reviews will be considered for peer review from qualified experts in the area. These manuscripts should be no longer than 6000 words and include no more than four figures and tables and 120 references.

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Consensus Statements may be submitted by professional societies. All such submission will be subjected to peer review, must be modifiable in response to criticisms, and will be published only if they meet the Journal's usual editorial standards. These manuscripts should typically be no longer than 4000 words and include no more than six figures and tables and 120 references.

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- All tables and figures must be placed after the text and must be labeled.
- Each section (abstract, text, references, tables, figures) should start on a separate page.

- Manuscripts should be prepared as word document (*.doc) or rich text format (*.rtf).

Title Page

The title page should include the following:

- Full title
- Authors' names and institutions.
- Short title of not more than 40 characters for page headings
- At least three and maximum eight key words. Do not use abbreviations in the key words
- Word count (excluding abstract, figure legends and references)
- Corresponding author's e-mail and post address, telephone and fax numbers
- Name and address of person to whom reprint requests should be addressed
- Any grants or fellowships supporting the writing of the paper
- The ORCID (Open Researcher and Contributor ID) number of the all authors should be provided while sending the manuscript. A free registration can be done at <http://orcid.org>.

Structured Abstracts (According to the The Journal of the American Medical Association)

Original Articles should be submitted with structured abstracts of no more than 250 words. All information reported in the abstract must appear in the manuscript. The abstract should not include references. Please use complete sentences for all sections of the abstract. Structured abstract should include background, objective, methods, results and conclusion.

What is already known on this topic?

What this study adds?

These two items must be completed before submission. Each item should include at most 2-3 sentences and at most 50 words focusing on what is known and what this study adds.

Review papers do not need to include these boxes.

Introduction

The article should begin with a brief introduction stating why the study was undertaken within the context of previous reports.

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All clinical investigations described in submitted manuscripts must have been conducted in accordance with the guidelines in the Declaration of Helsinki and has been formally approved by the appropriate institutional review committees. All manuscripts must indicate that such approval was obtained and that informed consent was obtained from subjects in all experiments involving humans. The study populations should be described in detail. Subjects must be identified only by number or letter, not by initials or names. Photographs of patients' faces should be included only if scientifically relevant. Authors must obtain written consent from the patient for use of such photographs.

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For clinical trial reports to be considered for publication in the Journal, prospective registration, as endorsed by the International Conference of Medical Journal Editors, is required. We recommend use of <http://www.clinicaltrials.gov>.

Experimental Animals

A statement confirming that all animal experimentation described in the submitted manuscript was conducted in accord with accepted standards of

humane animal care, according to the Declaration of Helsinki and Genova Convention, should be included in the manuscript.

Materials and Methods

These should be described and referenced in sufficient detail for other investigators to repeat the work. Ethical consent should be included as stated above.

The name of the ethical committee, approval number should be stated.

Results

The Results section should briefly present the experimental data in text, tables, and/or figures. Do not compare your observations with that of others in the results section.

Discussion

The Discussion should focus on the interpretation and significance of the findings with concise objective comments that describe their relation to other work in that area and contain study limitations.

Study Limitations

Limitations of the study should be detailed. In addition, an evaluation of the implications of the obtained findings/results for future research should be outlined.

Conclusion

The conclusion of the study should be highlighted.

Acknowledgments (Not Required for Submission)

An acknowledgment is given for contributors who may not be listed as authors, or for grant support of the research.

Authorship Contribution

The kind of contribution of each author should be stated.

References

References to the literature should be cited in numerical order (in parentheses) in the text and listed in the same numerical order at the end of the manuscript on a separate page or pages. The author is responsible for the accuracy of references.

Number of References: Case Report max 30 / Original Articles max 50

Examples of the reference style are given below. Further examples will be found in the articles describing the Uniform Requirements for Manuscripts Submitted to Biomedical Journals (Ann Intern Med.1988; 208:258-265, Br Med J. 1988; 296:401-405). The titles of journals should be abbreviated according to the style used in the Index Medicus.

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Books: List all authors or editors.

Sample References

Papers Published in Periodical Journals: Gungor N, Saad R, Janosky J, Arslanian S. Validation of surrogate estimates of insulin sensitivity and insulin secretion in children and adolescents. J Pediatr 2004;144:47-55.

Papers Only Published with DOI Numbers: Knops NB, Sneeuw KC, Brand R, Hile ET, de Ouden AL, Wit JM, Verloove-Vanhorick SP. Catch-up growth up to ten years of age in children born very preterm or with very low birth weight. BMC Pediatrics 2005 doi: 10.1186/1471-2431-5-26.

Book Chapters: Darendeliler F. Growth Hormone Treatment in Rare Disorders: The KIGS Experience. In: Ranke MB, Price DA, Reiter EO (eds). Growth Hormone Therapy in Pediatrics: 20 Years of KIGS. Basel, Karger, 2007;213-239.

Books: Practical Endocrinology and Diabetes in Children. Raine JE, Donaldson MDC, Gregory JW, Savage MO. London, Blackwell Science, 2001;37-60.

Tables

Tables must be constructed as simply as possible. Each table must have a concise heading and should be submitted on a separate page. Tables must not simply duplicate the text or figures. Number all tables in the order of their citation in the text. Include a title for each table (a brief phrase, preferably no longer than 10 to 15 words). Include all tables in a single file following the manuscript.

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