

Combined Orthodontic-Prosthodontic Digital Workflow for Management of *TP63*-Related Ectodermal Dysplasia

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Introduction

Tumor protein p63 is a transcription factor encoded by *TP63* gene that plays a vital role in the development and maintenance of the skin, ectodermal tissues, orofacial structures, and limbs. Pathologic variants in different domains of *TP63* result in different ectodermal dysplasia syndromes with overlapping features.

Acro-dermo-ungual-lacrimal-tooth (ADULT) syndrome is a specific *TP63*-related disorder characterized by:

- Ectodermal dysplasia affecting the teeth, hair, nails, and sweat glands
- Syndactyly of the hands and feet
- Excessive freckling¹

There is a paucity of literature in the dental management of complex syndromic patients with skeletal, dental, or alveolar deficiencies, making the treatment planning process challenging.

Patient presentation

A 15-year-old male diagnosed with ADULT syndrome presents to the Prosthodontics Department at the University of Rochester. The patient was referred by the Orthodontics Department for a restorative evaluation prior to orthodontic therapy.

Clinical examination reveals classic features of ADULT syndrome, including:

- Ectodermal dysplasia
- Syndactyly of the hands and feet
- Excessive freckling



A) Frontal view B) Profile view C) Right foot radiograph D) Retracted intraoral

Essential aspects of interdisciplinary collaboration

In this case, early collaboration between the Prosthodontics and Orthodontics Departments is essential to:

- Establish a prosthetically-driven plan to guide tooth positioning and space management
- Strategically maintain primary teeth to preserve bone for future implant placement
- Integrate a digital wax-up and face scan for virtual mock-up and orthodontic planning
- Define the treatment timeline and sequence of care
- Set clear expectations with the patient and family through visual simulation of treatment goals

Digital treatment planning workflow

A digital workflow by Papaspyridakos et al. (2022) was adapted to guide interdisciplinary treatment planning.²

Data acquisition

Diagnostics

- Incisal edge position, OVD, centric relation (CR)

Dental Findings

Prosthodontics	Orthodontics
• Oligodontia	• Severe deep bite
• Retained primary teeth	• Midline discrepancy
• Abnormal tooth morphology	• Skeletal class I with horizontal growth pattern
• Excessive incisal display	• Class II dental malocclusion
• Retroclined #8,9	• Crossbite of UR-E with LR-E
• Severe wear #M-P	
• Loss of OVD	
• Generalized spacing	

Photography

- Intraoral and extraoral photographs

Intraoral scan

- Intraoral scan at proposed OVD recorded in CR



Extraoral scan

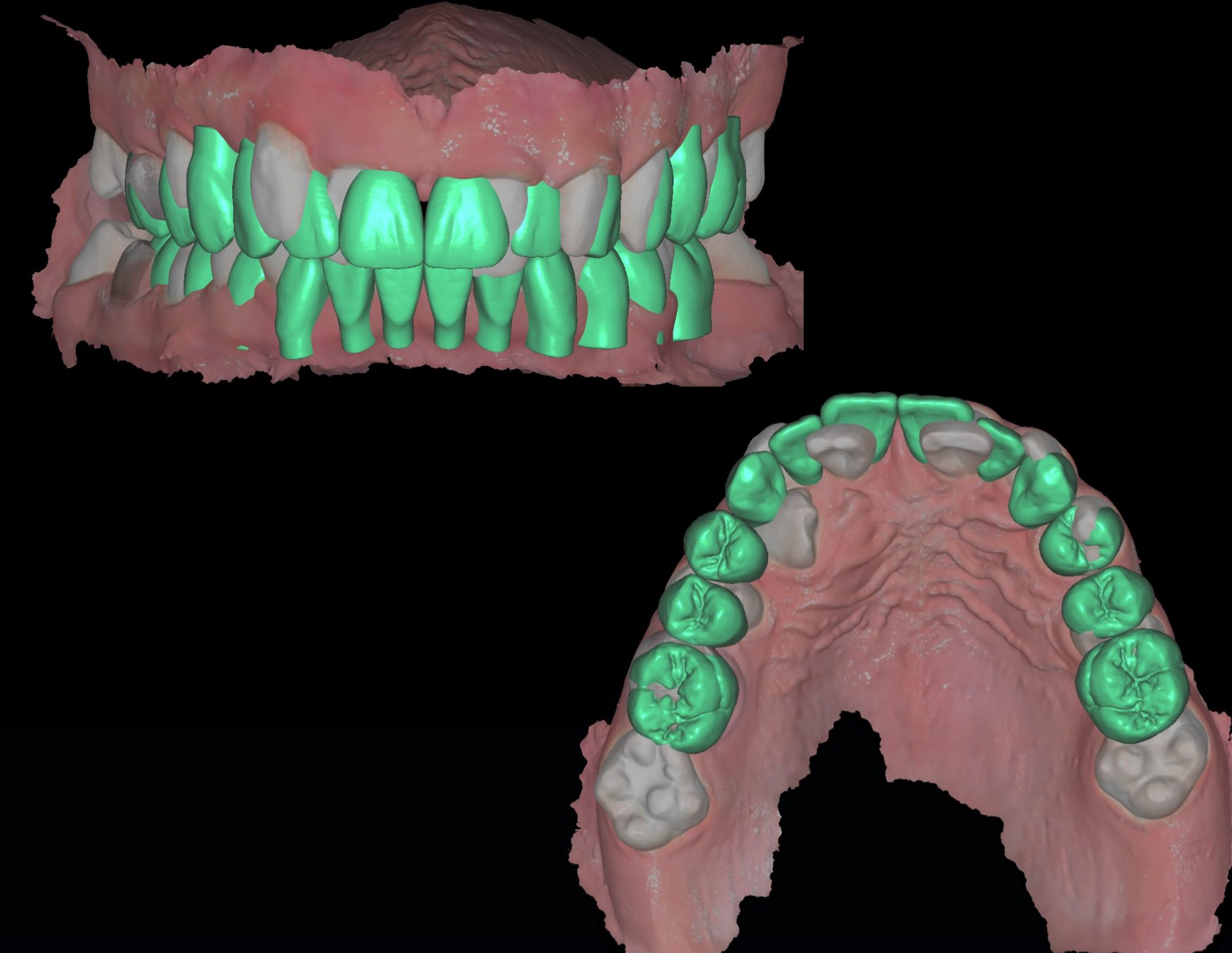
- Facial scan for digital smile design



Computer-aided design

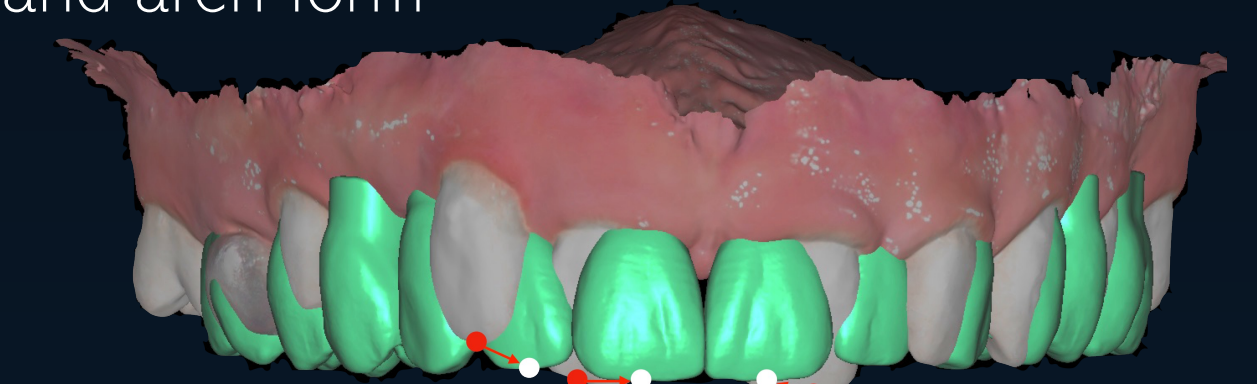
Data merging

- Intraoral scan, facial scan, and photographs merged in computer-aided design software
- Digital wax-up at proposed OVD



Digital orthodontic planning

- Virtual mapping of ideal tooth movements
- Visualize size discrepancies between natural teeth and future crowns
- Prosthetically-driven plan to guide tooth movement:
 - Intrude and procline #8,9
 - Maintain primary teeth for future implant placement
 - Expand arch form



Conclusions

- In complex syndromic cases, clinical mock-ups are often not feasible due to significant changes in anticipated tooth positioning
- Incorporating an extraoral facial scan allows for a true 3D virtual mock-up, allowing for patient education and visualization
- Virtual mock-up allows providers to simulate restorative outcomes early and align orthodontic tooth movements accordingly
- Early prosthodontic involvement is essential to avoid premature extractions and to guide space management for future restorative needs
- For patients undergoing treatment over several years, especially those affected by self-esteem challenges, visualizing the proposed outcome provides motivation, reassurance, and a clearer understanding of their treatment

References

1. Brunner, H. G., Hamel, B. C. J., & van Bokhoven H. (2002). P63 gene mutations and human developmental syndromes. *American Journal of Medical Genetics*, 112(3), 284–290. <https://doi.org/10.1002/ajmg.10778>
2. Papaspyridakos, P., Bedrossian, A., De Souza, A., Bokhary, A., Gonzaga, L., & Chochlidakis, K. (2022). Digital workflow in implant treatment planning for terminal dentition patients. *Journal of Prosthodontics*, 31(6), 543–548. <https://doi.org/10.1111/jopr.13510>