



Unlocking the Future: Genetic Testing's Pivotal Role in Pediatric Craniofacial Plastic Surgery



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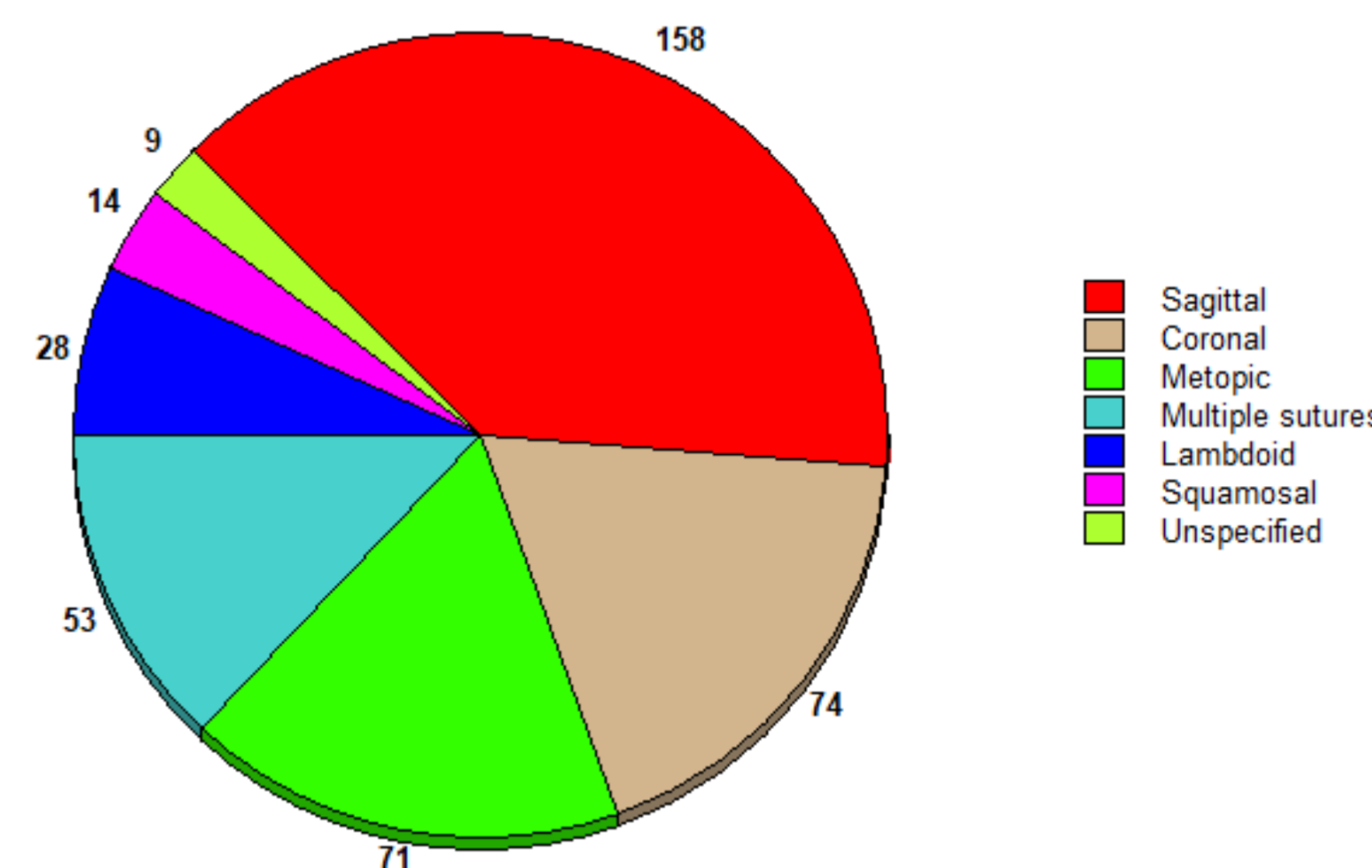
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Background

- Craniofacial anomalies, including craniosynostosis, represent a diverse group of congenital conditions characterized by abnormalities in the skull and facial structures.
- Genetic testing has emerged as a revolutionary adjunct to traditional diagnostic approaches, offering insights into the underlying genetic factors contributing to craniofacial abnormalities.
- Craniosynostosis is a condition where the sutures in a child's skull close too early, affecting about 1 in 2,500 born.
- The disorder is recognized as having a complex and multifactorial origin, with genetic, epigenetic, and environmental variables thought to contribute to its development.
- Missed early opportunities for surgical and behavioral intervention result in a worsened prognosis of those affected.
- There is a critical need to assess the potential role of genetic testing in pediatric Plastic Surgery practice.
- We performed a cursory proof-of-principle in patients diagnosed with craniosynostosis at our institution.

Methods and Overview



A retrospective review was performed of patients who presented to a single institution craniosynostosis evaluation between January 1997 and December 2020.

Results

- 356 out of 719 CS patients (49.5%) were referred for genetic counseling
- 280 patients received some form of genetic testing (48.7%)
- 252 patients had notable findings in their testing (90%)
- 89 patients (12.4%) had a sibling with a known genetic anomaly; 15.9% an affected mother, 15.3% an affected father, 25.7 % an affected maternal grandparent, 18.8% an affected paternal grandparent, 25.3 % other affected family members
- 29 patients (10.4% of 280 patients that were tested) had consanguineous parents
- Of the 280 that received genetic testing, 61.1% were male and 38.9% female at birth.
- The majority of patients w/ a recorded race were white (120 total).
- Common recorded religions were 29 (Catholic), 11 (Jewish), 110 (No religion), 85 (Other)

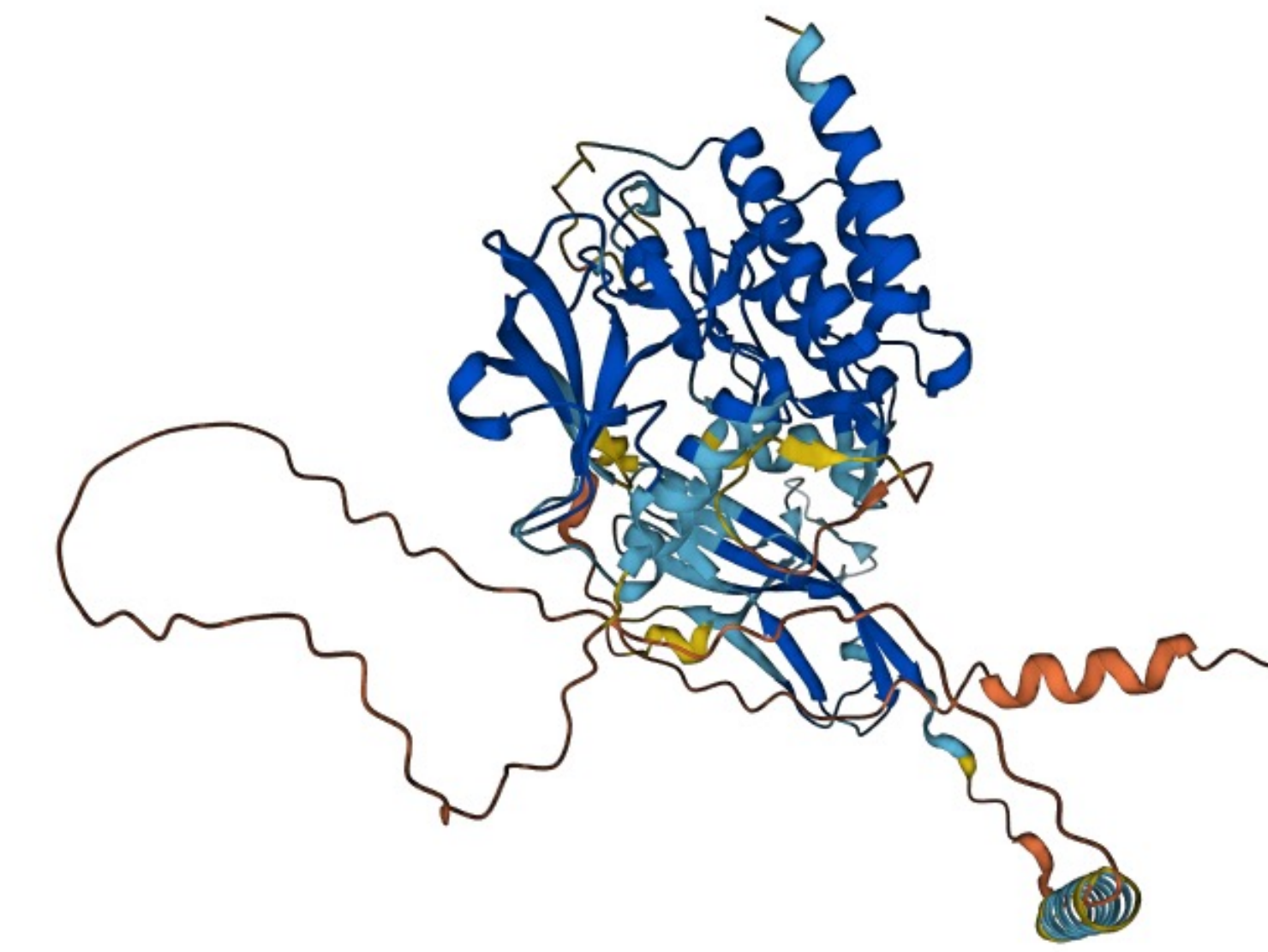


Figure 1. Physiologic FGFR2, 3D view.

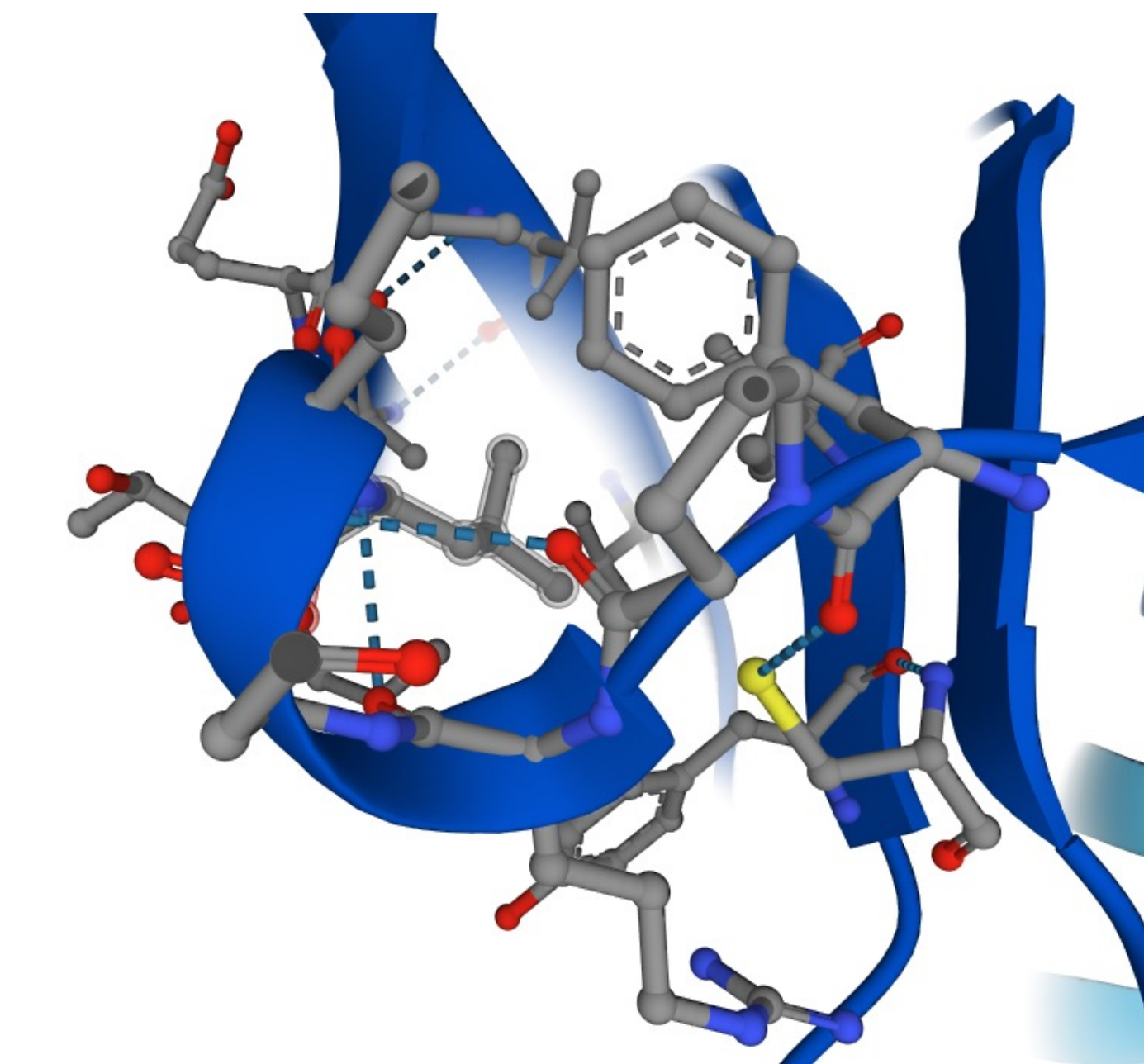


Figure 2. FGFR2, p.Pro253 Arg.

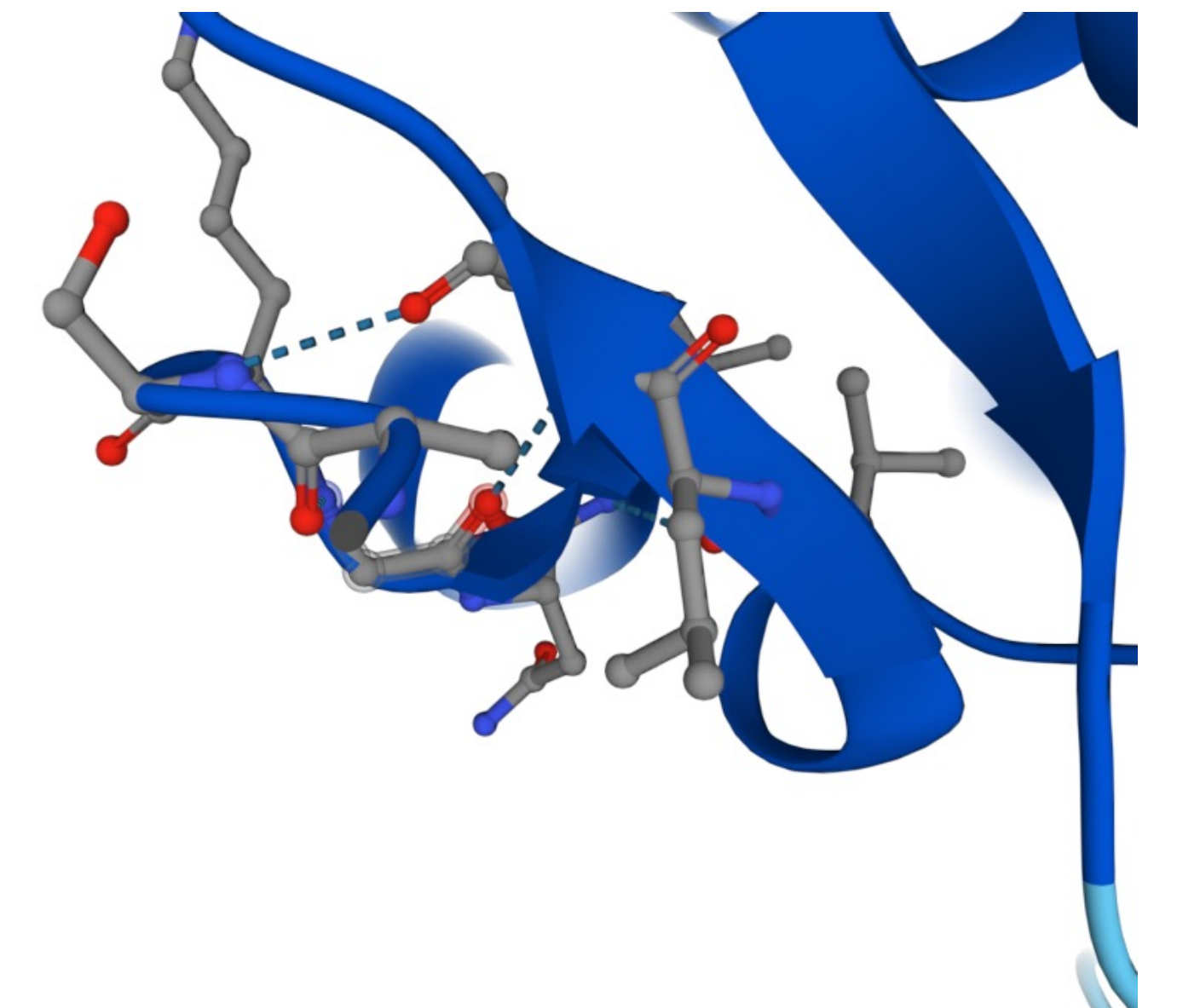


Figure 3. FGFR2, p. Cys342Tyr.

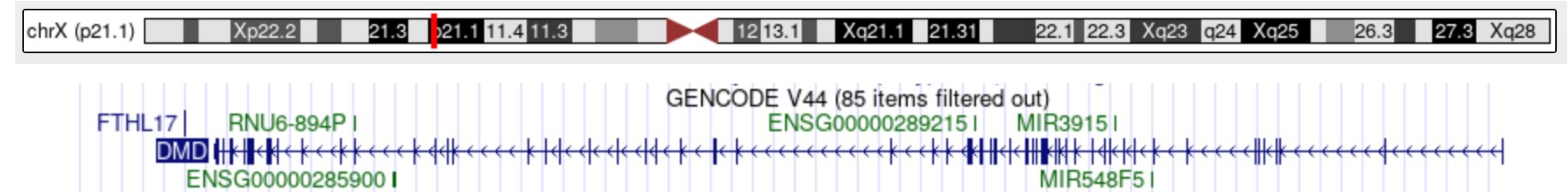
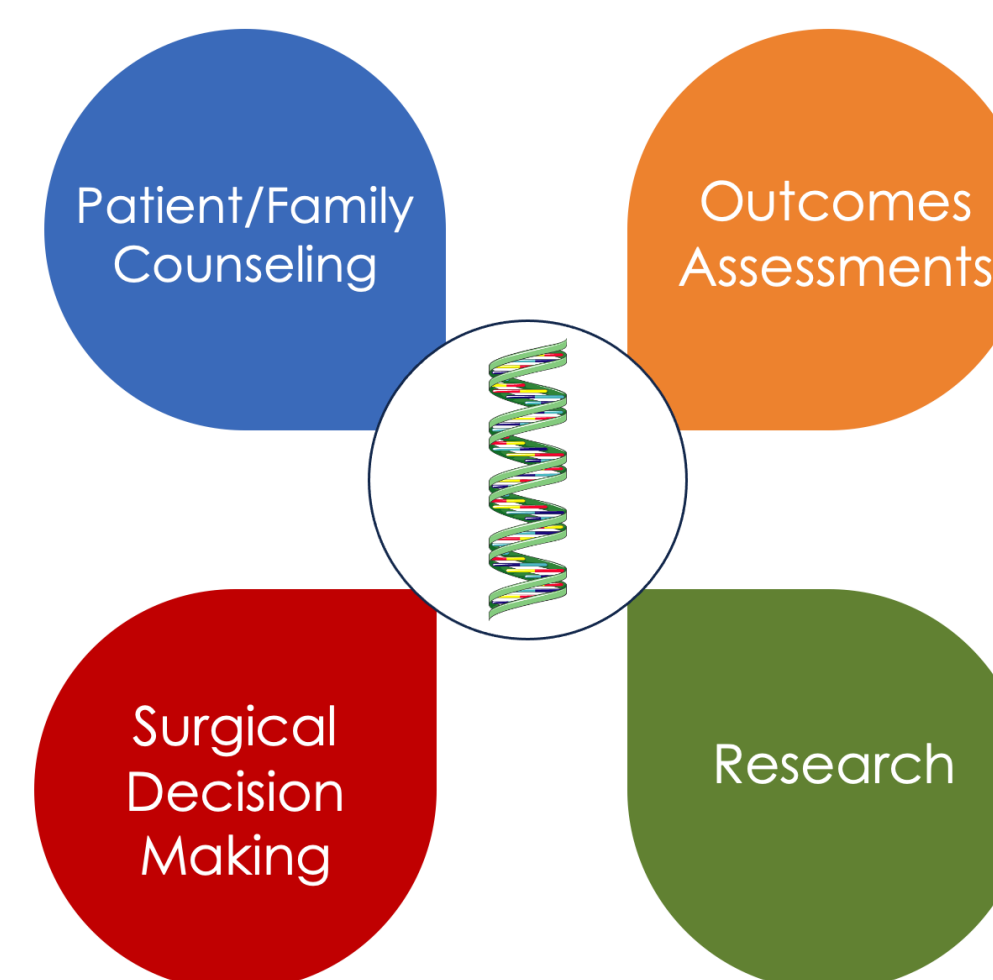


Figure 4. SNP microarray demonstrated an XP21.1 deletion including exons 45-52 of *DMD*.

Conclusions



- Genetic testing not only aids in identifying the etiology of craniofacial anomalies but also plays a crucial role in guiding treatment decisions, informing surgical planning, and predicting long-term outcomes.
- Genetic referrals and evaluations remain underutilized in clinical practice despite improvements in cost.
- Standardized testing enables clinicians to offer personalized management strategies tailored to the unique genetic profiles and clinical presentations of individual patients. The future of pediatric craniofacial plastic surgery, particularly in complex conditions like, has much to gain from the integration of genetic testing in standard clinical practice.
- Future studies should examine the cost-effectiveness and clinical merits of the integration of standardized genetic testing in pediatric craniofacial Plastic Surgery.

