GENETICS CENTER

Comprehensive FGFR and TWIST-related Craniosynostosis Syndrome Test Panel

TEST	AREAS EVALUATED	METHOD	CPT's
Craniosynostosis Comprehensive Panel (FGFR1, FGFR2, FGFR3 and TWIST)	**FGFR1**: Pro252Arg **FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17 **FGFR3**: Pro250Arg and exon 10 **TWIST**: Exon1	RFLP and Sequencing 11 exons	81401, 81406, G0452
Apert Syndrome	**FGFR2**: Exon8	Sequencing 1 exon	81403, G0452
Crouzon Syndrome	**FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17	Sequencing 9 exons	81405, G0452
Testing for Known Familial Mutation	**FGFR1**, **FGFR2**, **FGFR3** or **TWIST** select exon analysis	Sequencing 1 exon	81403, G0452
Crouzon Syndrome (including CS Acanthosis Nigracans)	**FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17 **FGFR3**: Exon 10	Sequencing 10 exons	81405, G0452
FGFR1	**FGFR1**: Pro252Arg	RFLP	81400, G0452
FGFR2	**FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17	Sequencing 9 exons	81405, G0452
FGFR3	**FGFR3**: Pro250Arg and Exon 10	RFLP and Sequencing 1 exon	81401, 81403, G0452
Muenke Syndrome	**FGFR3**: Pro250Arg	RFLP	81400, G0452
Isolated Coronal Craniosynostosis (including Muenke Syndrome)	**FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17 **FGFR3**: Pro250Arg	RFLP and Sequencing 9 exons	81400, 81405, G0452
Pfeiffer Syndrome	**FGFR1**: Pro252Arg **FGFR2**: Exons 3,5,8,10,11, 14, 15, 16, 17	RFLP and Sequencing 9 exons	81400, 81405, G0452
Saethre-Chotzen Syndrome	**TWIST**: Exon1	Sequencing 1 exon	81403, G0452

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