17. SKELETAL				
	a	**Skeletal Dysplasias**		
	MACH	Achondroplasia mutation study	4 ml blood EDTA	3-4 wks
	MACS	Achondroplasia Gene Sequencing FGFR3	4 ml blood EDTA	6-8 wks
	MSAD	Achondroplasia with Acantharis Nigricans - SADDAN mutation Lys 650 Met in FGFR3 gene	4 ml blood EDTA	3-4 wks
	MEMG1	Bowen Conradi Syndrome-EMG1 sequencing	4ml EDTA	4 to 6 wks
	MTGFB1	Camurati-Engelmann disease - TGFβ-1 gene sequencing	4 ml blood EDTA	6-8 wks
	MXLCDP	Chondrodysplasia Punctata X linked CDP1-ARSE gene sequencing	4 ml EDTA	4 to 6 wks
	MRUNX2	Cleidocranial dysplasia - RUNX2 gene sequencing	4 ml blood EDTA	3-4 wks
	MEBP	Conradi Hunermann synd. EBP gene sequencing	4 ml blood EDTA	6-8 wks
	MSLC26 A2	Diastrophic Dysplasia-SLC26A2 gene sequencing	4 ml blood EDTA	2-3 wks
	MCANT	Desbuquois Dysplasia- CANT1 gene sequencing	4 ml EDTA	3 to 4 wks
	MCHST	Desbuquois Dysplasia- CHST3 gene sequencing	4 ml EDTA	3 to 4 wks
	MACVR1	Fibrodysplasia ossificans progressiva (FOP)- ACVR1 gene sequencing	4 ml EDTA	3 to 4 wks
	MGDF5	Grebe syndrome- GDF5 gene sequencing	4 ml EDTA	3 to 4 wks
	MHCM	Hypochondroplasia common mutation	4 ml blood EDTA	3-4 wks
	MHCS	Hypochondroplasia (Sequencing)	4 ml blood EDTA	6-8 wks
	MMESP2	Jarcho Levin syndrome - MESP2 gene sequencing	4 ml blood EDTA	6-8 wks
	MSHOX	Leri-Weill dyschondrosteosis and Langer mesomelic dysplasia SHOX gene sequencing	4 ml EDTA	3 to 4 wks
	MC10S	Metaphyseal Dysplasia - COL 10A Gene Sequencing	4 ml blood EDTA	6-8 wks
	MOI	Osteogenesis imperfecta-COL1A1 known mutation analysis	4 ml blood EDTA	3-4 wks
	MTCIRG	Osteopetrosis TCIRG gene common mutations (exon 13 & 18)	4 ml EDTA	2 to 3 wks
	MCOMP	Pseudoachondroplasia - COMP gene sequencing of exons 10-14, 16	4 ml blood EDTA	3-4 wks
	MCTSK	Pyknodysostosis-CTSK sequencing	4ml EDTA blood	4 to 6 wks
	MTBCE	Sanjad Sakati syndrome/common deletion - TBCE gene	4 ml blood EDTA	3-4 wks

	MSEDS	Spondyloepiphyseal Dysplasia X-Linked gene Sequencing	4 ml blood EDTA	3-4 wks
	MKIF22	Spondyloepimetaphaseal Leptodactylic type dysplaisa/SED - KIF22 exon 4	4 ml EDTA	2 to 3 wks
	MTD	Thanatophoric dwarfism (common mutation)	4 ml blood EDTA	3-4 wks
	MTDS	Thanatophoric Dwarfism - Sequencing	4 ml blood EDTA	3-4 wks
	MFGS	FGFR3 Gene Sequencing (Achondroplasia , Hypochondroplasia Thanatophoric Dwarfism)	4 ml blood EDTA	6-8 wks
	b	**Craniosynostosis Syndromes**		
	MAPE	Apert syndrome mutation study	4 ml blood EDTA	3-4 wks
	MCRZ	Crouzon Disease : FGFR 2 Mutation (SER 35 Y CYS)	4 ml blood EDTA	3-4 wks
	MFGFR1S	FGFR1 gene sequencing	4 ml blood EDTA	6-8 wks
	MFGFR2S	FGFR2 sequencing (Crouzon, Jackson Weiss, Pfeiffer, AD craniosynostosis)	4 ml blood EDTA	6-8 wks
	MCNS	Craniosynostosis (non specific)	4 ml blood EDTA	3-4 wks
	MTWISTD	Saethre- Chotzen syndrome - TWIST Dosage analysis	4ml EDTA	2 to 3 wks
	MTWISTS	Saethre- Chotzen syndrome - TWIST gene sequencing	4ml EDTA	2 to 3 wks
	MTWISTSD	Saethre- Chotzen syndrome - TWIST gene Dosage +sequencing	4ml EDTA	4 to 6 wks
	MRAB23	Carpenter syndrome- RAB23 gene sequencing	4 ml blood EDTA	6-8 wks