E	Dysmorphic Syndromes and Skeletal Dysplasias		
1	Molecular diagnosis of distal renal tubular acidosis	ATP6V1B1, ATP6V0A4, SLC4A1	
2	Molecular diagnosis of Leri – Weill syndrome	SHOX	
3	Molecular diagnosis of cardiofaciocutaneous syndrome	BRAF, HRAS, KRAS	
4	Molecular diagnosis of Costello syndrome	BRAF, HRAS, KRAS	
5	Molecular diagnosis of Stickler syndrome	COL2A1, COL11A1	
6	Molecular diagnosis of Spondyloepiphyseal Dysplasia	COL2A1	
7	Molecular diagnosis of Achondrogenesis type 2	COL2A1	
8	Molecular diagnosis of Kniest dysplasia	COL2A1	
9	Molecular diagnosis of Muenke syndrome	COL2A1	
10	Molecular diagnosis of Apert syndrome	FGFR2	
11	Molecular diagnosis for Crouzon disease	FGFR2	
12	Molecular diagnosis of Pfeiffer syndrome	FGFR1, FGFR2	
13	Molecular diagnosis of Saethre – Chotzen syndrome	TWIST	
14	Molecular diagnosis of Achondroplasia	FGFR3	
15	Molecular diagnosis of Hypochondroplasis	FGFR3	
16	Molecular diagnosis of Craniofrontonasal Dysplasia	EFNB1	
17	Molecular diagnosis of Anhidrotic Ectodermal Dysplasia	ED1, EDAR, EDARADD	
18	Molecular diagnosis of Li – Fraumeni syndrome	P53 Gene	