

Supplement Table1 EXT1's variant data

gene	lovd id	type	variant on genome	clinical classification
EXT1	130513	substitution	g.118842473A>C	VUS
EXT1	167144	substitution	g.118849345A>G	likely pathogenic
EXT1	170458	deletion	g.119122585del	pathogenic
EXT1	194531	deletion	g. (?_118811602)_ (119124058_?) del	likely pathogenic
EXT1	194532	deletion	g. (?_118811602)_ (119124058_?) del	likely pathogenic
EXT1	194533	deletion	g. (?_118811602)_ (119124058_?) del	likely pathogenic
EXT1	194534	deletion	g. (?_118811602)_ (119124058_?) del	likely pathogenic
EXT1	194535	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194536	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194537	deletion	g. (118842589_118847682)_ (118849441_119122323) del	likely pathogenic
EXT1	194538	deletion	g. (118842589_118847682)_ (118849441_119122323) del	likely pathogenic
EXT1	194539	deletion	g. (?_118811602)_ (118849441_119122323) del	likely pathogenic
EXT1	194540	deletion	g. (?_118811602)_ (118849441_119122323) del	likely pathogenic
EXT1	194541	deletion	g. (118825201_118830673)_ (118832034_118834703) del	likely pathogenic
EXT1	194542	deletion	g. (118819617_118825110)_ (118825201_118830673) del	likely pathogenic
EXT1	194546	substitution	g.119124061A>G	benign
EXT1	194547	substitution	g.119124569C>G	benign
EXT1	194548	substitution	g.119124443C>G	benign
EXT1	194549	substitution	g.119124350C>T	benign
EXT1	194550	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194551	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194552	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194553	deletion	g. (118842589_118847682)_ (119124058_?) del	likely pathogenic
EXT1	194554	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194555	deletion	g. (118849441_119122323)_ (119124058_?) del	likely pathogenic
EXT1	194556	deletion	g. (?_118811602)_ (119124058_?) del	pathogenic
EXT1	194557	deletion	g. (?_118811602)_ (119124058_?) del	likely pathogenic

EXT1	194558	deletion	g. (?_118811602)_(118812137_118816960)del	likely pathogenic
EXT1	194559	deletion	g. (?_118811602)_(118849441_119122323)del	likely pathogenic
EXT1	194560	substitution	g. 119123285T>C	likely pathogenic
EXT1	194561	substitution	g. 119123285T>A	likely pathogenic
EXT1	194562	substitution	g. 119123284A>T	likely pathogenic
EXT1	194563	substitution	g. 119123284A>T	likely pathogenic
EXT1	194564	substitution	g. 119123282G>A	pathogenic
EXT1	194565	duplication	g. 119123276dup	likely pathogenic
EXT1	194566	duplication	g. 119123276dup	likely pathogenic
EXT1	194567	deletion	g. 119123259del	likely pathogenic
EXT1	194568	deletion	g. 119123249del	likely pathogenic
EXT1	194569	deletion	g. 119123246del	likely pathogenic
EXT1	194570	deletion	g. 119123240_119123241del	likely pathogenic
EXT1	194571	deletion	g. 119123240_119123241del	likely pathogenic
EXT1	194572	duplication	g. 119123236dup	likely pathogenic
EXT1	194573	deletion	g. 119123225del	likely pathogenic
EXT1	194574	deletion	g. 119123220del	likely pathogenic
EXT1	194575	deletion	g. 119123202_119123206del	likely pathogenic
EXT1	194576	deletion	g. 119123200del	likely pathogenic
EXT1	194577	deletion	g. 119123200del	likely pathogenic
EXT1	194578	deletion	g. 119123194del	likely pathogenic
EXT1	194579	insertion	g. 119123171_119123172insAA	likely pathogenic
EXT1	194580	deletion	g. 119123171del	likely pathogenic
EXT1	194581	substitution	g. 119123171C>A	likely pathogenic
EXT1	194582	substitution	g. 119123171C>A	likely pathogenic
EXT1	194583	substitution	g. 119123169T>C	benign
EXT1	194584	deletion	g. 119123168del	likely pathogenic
EXT1	194585	deletion	g. 119123168_119123169del	likely pathogenic
EXT1	194586	substitution	g. 119123164C>T	benign
EXT1	194587	insertion	g. 119123148_119123149insGGGGTGC	likely pathogenic
EXT1	194588	deletion	g. 119123145del	likely pathogenic
EXT1	194589	deletion	g. 119123112del	likely pathogenic
EXT1	194590	deletion	g. 119123106del	likely pathogenic
EXT1	194591	deletion	g. 119123104del	likely pathogenic
EXT1	194592	deletion	g. 119123104del	likely pathogenic
EXT1	194593	insertion	g. 119123097_119123098insA	likely pathogenic
EXT1	194594	deletion	g. 119123090del	likely pathogenic
EXT1	194595	substitution	g. 119123082C>T	likely pathogenic
EXT1	194596	substitution	g. 119123082C>T	likely pathogenic
EXT1	194597	substitution	g. 119123072C>A	likely pathogenic
EXT1	194598	substitution	g. 119123072C>T	likely pathogenic
EXT1	194599	deletion	g. 119123071del	likely pathogenic
EXT1	194600	deletion	g. 119123071del	likely pathogenic
EXT1	194601	deletion	g. 119123048del	likely pathogenic
EXT1	194602	deletion	g. 119123048del	likely pathogenic

EXT1	194603	deletion	g.119123047del	likely pathogenic
EXT1	194604	deletion	g.119123044del	likely pathogenic
EXT1	194605	deletion	g.119123044del	likely pathogenic
EXT1	194606	deletion	g.119123044del	likely pathogenic
EXT1	194607	duplication	g.119123044dup	likely pathogenic
EXT1	194608	duplication	g.119123044dup	likely pathogenic
EXT1	194609	duplication	g.119123044dup	likely pathogenic
EXT1	194610	insertion	g.119123037_119123038 insG	likely pathogenic
EXT1	194611	deletion	g.119123038del	likely pathogenic
EXT1	194612	substitution	g.119123036G>A	likely pathogenic
EXT1	194613	substitution	g.119123036G>A	likely pathogenic
EXT1	194614	substitution	g.119123036G>A	likely pathogenic
EXT1	194615	deletion	g.119123006del	likely pathogenic
EXT1	194616	substitution	g.119122955T>A	likely pathogenic
EXT1	194617	deletion	g.119122935del	likely pathogenic
EXT1	194618	insertion	g.119122933_119122934 insG	likely pathogenic
EXT1	194619	substitution	g.119122929G>T	likely pathogenic
EXT1	194620	substitution	g.119122929G>T	likely pathogenic
EXT1	194621	substitution	g.119122929G>T	likely pathogenic
EXT1	194622	substitution	g.119122929G>C	likely pathogenic
EXT1	194623	substitution	g.119122922G>A	likely pathogenic
EXT1	194624	substitution	g.119122922G>A	likely pathogenic
EXT1	194625	substitution	g.119122922G>A	likely pathogenic
EXT1	194626	indel	g.119122917_119122918 delinsG	likely pathogenic
EXT1	194627	deletion	g.119122916_119122917 del	likely pathogenic
EXT1	194628	deletion	g.119122916_119122917 del	likely pathogenic
EXT1	194629	deletion	g.119122916_119122917 del	likely pathogenic
EXT1	194630	deletion	g.119122916_119122917 del	likely pathogenic
EXT1	194631	duplication	g.119122916dup	likely pathogenic
EXT1	194632	deletion	g.119122905del	likely pathogenic
EXT1	194633	deletion	g.119122897_119122898 del	likely pathogenic
EXT1	194634	deletion	g.119122891del	likely pathogenic
EXT1	194635	insertion	g.119122865_119122867 del	likely pathogenic
EXT1	194636	insertion	g.119122865_119122867 del	likely pathogenic
EXT1	194637	insertion	g.119122865_119122867 del	likely pathogenic
EXT1	194638	deletion	g.119122857del	likely pathogenic
EXT1	194639	deletion	g.119122850_119122854 del	likely pathogenic
EXT1	194640	deletion	g.119122830del	likely pathogenic
EXT1	194641	deletion	g.119122828_119122829 del	likely pathogenic
EXT1	194642	deletion	g.119122828_119122829 del	likely pathogenic

EXT1	194643	deletion	g. 119122825_119122826 del	likely pathogenic
EXT1	194644	deletion	g. 119122825_119122826 del	likely pathogenic
EXT1	194645	deletion	g. 119122825_119122826 del	likely pathogenic
EXT1	194646	deletion	g. 119122809_119122810 del	likely pathogenic
EXT1	194647	substitution	g. 119122804A>C	likely pathogenic
EXT1	194648	substitution	g. 119122796C>G	likely pathogenic
EXT1	194649	deletion	g. 119122798_119122799 del	likely pathogenic
EXT1	194650	substitution	g. 119122793G>A	likely pathogenic
EXT1	194651	substitution	g. 119122786G>T	likely pathogenic
EXT1	194652	deletion	g. 119122775_119122776 del	likely pathogenic
EXT1	194653	deletion	g. 119122771del	likely pathogenic
EXT1	194654	insertion	g. 119122769_119122770 insTTTT	likely pathogenic
EXT1	194655	deletion	g. 119122752_119122759 del	likely pathogenic
EXT1	194656	deletion	g. 119122757del	likely pathogenic
EXT1	194657	deletion	g. 119122757del	likely pathogenic
EXT1	194658	substitution	g. 119122751G>A	likely pathogenic
EXT1	194659	deletion	g. 119122749_119122750 del	likely pathogenic
EXT1	194660	deletion	g. 119122749_119122750 del	likely pathogenic
EXT1	194661	duplication	g. 119122737dup	likely pathogenic
EXT1	194662	deletion	g. 119122737_119122738 del	likely pathogenic
EXT1	194663	indel	g. 119122724_119122726 delinsG	likely pathogenic
EXT1	194664	deletion	g. 119122716del	likely pathogenic
EXT1	194665	substitution	g. 119122714A>G	likely pathogenic
EXT1	194666	substitution	g. 119122714A>C	likely pathogenic
EXT1	194667	deletion	g. 119122713_119122717 del	likely pathogenic
EXT1	194668	deletion	g. 119122704del	likely pathogenic
EXT1	194669	deletion	g. 119122696del	likely pathogenic
EXT1	194670	substitution	g. 119122694C>G	likely pathogenic
EXT1	194672	substitution	g. 119122687C>T	likely pathogenic
EXT1	194673	substitution	g. 119122687C>T	likely pathogenic
EXT1	194674	substitution	g. 119122686C>T	likely pathogenic
EXT1	194675	indel	g. 119122674_119122683 delinsC	likely pathogenic
EXT1	194676	substitution	g. 119122678T>C	likely pathogenic
EXT1	194677	duplication	g. 119122663_119122667 dup	likely pathogenic
EXT1	194678	deletion	g. 119122619_119122645 del	likely pathogenic
EXT1	194679	deletion	g. 119122623_119122643 del	likely pathogenic

EXT1	194680	deletion	g. 119122623_119122643 del	likely pathogenic
EXT1	194681	deletion	g. 119122623_119122643 del	likely pathogenic
EXT1	194682	indel	g. 119122620_119122641 delinsG	likely pathogenic
EXT1	194683	deletion	g. 119122622_119122635 del	likely pathogenic
EXT1	194684	indel	g. 119122622_119122635 delinsAAA	likely pathogenic
EXT1	194685	indel	g. 119122621_119122635 delinsAA	likely pathogenic
EXT1	194686	deletion	g. 119122627_119122635 del	likely pathogenic
EXT1	194687	substitution	g. 119122627C>T	likely pathogenic
EXT1	194688	duplication	g. 119122626dup	likely pathogenic
EXT1	194689	substitution	g. 119122607G>A	likely pathogenic
EXT1	194690	substitution	g. 119122607G>A	likely pathogenic
EXT1	194691	deletion	g. 119122608del	likely pathogenic
EXT1	194692	deletion	g. 119122608del	likely pathogenic
EXT1	194693	deletion	g. 119122606del	likely pathogenic
EXT1	194694	deletion	g. 119122569_119122583 del	likely pathogenic
EXT1	194695	substitution	g. 119122582G>C	likely pathogenic
EXT1	194696	deletion	g. 119122579_119122580 del	likely pathogenic
EXT1	194697	deletion	g. 119122577del	likely pathogenic
EXT1	194698	deletion	g. 119122577del	likely pathogenic
EXT1	194699	deletion	g. 119122573del	likely pathogenic
EXT1	194700	deletion	g. 119122573del	likely pathogenic
EXT1	194701	deletion	g. 119122573del	likely pathogenic
EXT1	194702	deletion	g. 119122573del	likely pathogenic
EXT1	194703	deletion	g. 119122562del	likely pathogenic
EXT1	194704	duplication	g. 119122562dup	VUS
EXT1	194705	deletion	g. 119122555del	likely pathogenic
EXT1	194706	deletion	g. 119122544del	likely pathogenic
EXT1	194707	deletion	g. 119122546_119122547 del	likely pathogenic
EXT1	194708	insertion	g. 119122543_119122544 insAA	likely pathogenic
EXT1	194709	deletion	g. 119122538del	likely pathogenic
EXT1	194710	substitution	g. 119122500G>T	likely pathogenic
EXT1	194711	substitution	g. 119122500G>C	likely pathogenic
EXT1	194712	substitution	g. 119122500G>C	likely pathogenic
EXT1	194713	deletion	g. 119122492_119122499 del	likely pathogenic
EXT1	194714	deletion	g. 119122494del	likely pathogenic
EXT1	194715	substitution	g. 119122484C>T	likely pathogenic
EXT1	194716	substitution	g. 119122484C>T	likely pathogenic
EXT1	194717	substitution	g. 119122484C>G	likely pathogenic
EXT1	194718	deletion	g. 119122484_119122485 del	likely pathogenic
EXT1	194719	insertion	g. 119122482_119122483 insA	likely pathogenic

EXT1	194720	deletion	g. 119122485del	likely pathogenic
EXT1	194721	deletion	g. 119122485del	likely pathogenic
EXT1	194722	deletion	g. 119122485del	likely pathogenic
EXT1	194723	indel	g. 119122482delinsAAGA	likely pathogenic
EXT1	194724	substitution	g. 119122478T>A	likely pathogenic
EXT1	194725	substitution	g. 119122475A>T	likely pathogenic
EXT1	194726	substitution	g. 119122475A>G	likely pathogenic
EXT1	194727	insertion	g. 119122474_119122475 insCT	likely pathogenic
EXT1	194728	substitution	g. 119122474T>C	likely pathogenic
EXT1	194729	substitution	g. 119122473G>C	likely pathogenic
EXT1	194730	deletion	g. 119122465_119122466 del	likely pathogenic
EXT1	194731	deletion	g. 119122466del	likely pathogenic
EXT1	194732	deletion	g. 119122466del	likely pathogenic
EXT1	194733	substitution	g. 119122456G>T	likely pathogenic
EXT1	194734	substitution	g. 119122456G>C	likely pathogenic
EXT1	194735	deletion	g. 119122452del	likely pathogenic
EXT1	194736	duplication	g. 119122450dup	likely pathogenic
EXT1	194737	substitution	g. 119122448T>C	likely pathogenic
EXT1	194738	substitution	g. 119122448T>C	likely pathogenic
EXT1	194739	substitution	g. 119122447C>G	likely pathogenic
EXT1	194740	substitution	g. 119122447C>A	likely pathogenic
EXT1	194741	substitution	g. 119122446C>G	likely pathogenic
EXT1	194742	substitution	g. 119122446C>G	likely pathogenic
EXT1	194743	substitution	g. 119122438A>T	pathogenic
EXT1	194744	duplication	g. 119122437_119122438 dup	likely pathogenic
EXT1	194745	duplication	g. 119122434dup	likely pathogenic
EXT1	194746	deletion	g. 119122420_119122421 del	likely pathogenic
EXT1	194747	duplication	g. 119122411dup	likely pathogenic
EXT1	194748	substitution	g. 119122405A>C	likely pathogenic
EXT1	194749	substitution	g. 119122392G>T	likely pathogenic
EXT1	194750	deletion	g. 119122385del	likely pathogenic
EXT1	194751	deletion	g. 119122382del	likely pathogenic
EXT1	194752	substitution	g. 119122374C>T	likely pathogenic
EXT1	194753	substitution	g. 119122373G>A	likely pathogenic
EXT1	194754	substitution	g. 119122370T>A	likely pathogenic
EXT1	194755	deletion	g. 119122352del	likely pathogenic
EXT1	194756	deletion	g. 119122345_119122346 del	likely pathogenic
EXT1	194757	deletion	g. 119122345_119122346 del	likely pathogenic
EXT1	194758	substitution	g. 119122339T>C	likely pathogenic
EXT1	194759	substitution	g. 119122328C>A	likely pathogenic
EXT1	194760	substitution	g. 119122323C>T	likely pathogenic
EXT1	194761	substitution	g. 119122323C>G	likely pathogenic
EXT1	194762	substitution	g. 119122323C>G	likely pathogenic
EXT1	194763	substitution	g. 119122323C>A	likely pathogenic
EXT1	194764	deletion	g. 119122322_119122325 del	likely pathogenic
EXT1	194765	substitution	g. 119122322A>G	likely pathogenic

EXT1	194766	deletion	g. 119122322_119122325 del	likely pathogenic
EXT1	194767	deletion	g. 119122315_119122318 del	VUS
EXT1	194768	substitution	g. 118849442T>C	likely pathogenic
EXT1	194769	substitution	g. 118849442T>C	likely pathogenic
EXT1	194770	substitution	g. 118849442T>C	likely pathogenic
EXT1	194771	substitution	g. 118849442T>C	likely pathogenic
EXT1	194772	substitution	g. 118849442T>C	likely pathogenic
EXT1	194773	substitution	g. 118849441C>T	likely pathogenic
EXT1	194774	substitution	g. 118849441C>T	likely pathogenic
EXT1	194775	substitution	g. 118849441C>A	likely pathogenic
EXT1	194776	deletion	g. (?_118811602)_(1188 49441_119122323)del	likely pathogenic
EXT1	194777	substitution	g. 118849437A>T	likely pathogenic
EXT1	194778	substitution	g. 118849437A>C	likely pathogenic
EXT1	194779	deletion	g. 118849429del	likely pathogenic
EXT1	194780	substitution	g. 118849427C>A	likely pathogenic
EXT1	194781	substitution	g. 118849427C>A	likely pathogenic
EXT1	194782	substitution	g. 118849411G>T	likely pathogenic
EXT1	194783	substitution	g. 118849411G>T	likely pathogenic
EXT1	194784	substitution	g. 118849411G>T	likely pathogenic
EXT1	194785	substitution	g. 118849411G>A	likely pathogenic
EXT1	194786	substitution	g. 118849399A>C	likely pathogenic
EXT1	194787	deletion	g. 118849398del	likely pathogenic
EXT1	194788	deletion	g. 118849391del	likely pathogenic
EXT1	194789	substitution	g. 118849387C>T	likely pathogenic
EXT1	194791	substitution	g. 118849387C>A	likely pathogenic
EXT1	194792	substitution	g. 118849385G>T	likely pathogenic
EXT1	194793	substitution	g. 118849385G>A	likely pathogenic
EXT1	194794	substitution	g. 118849385G>A	likely pathogenic
EXT1	194795	substitution	g. 118849385G>A	likely pathogenic
EXT1	194796	substitution	g. 118849385G>A	likely pathogenic
EXT1	194797	substitution	g. 118849385G>A	likely pathogenic
EXT1	194798	substitution	g. 118849385G>A	likely pathogenic
EXT1	194799	substitution	g. 118849385G>A	likely pathogenic
EXT1	194800	substitution	g. 118849385G>A	likely pathogenic
EXT1	194801	substitution	g. 118849385G>A	likely pathogenic
EXT1	194802	substitution	g. 118849385G>A	likely pathogenic
EXT1	194803	substitution	g. 118849385G>A	likely pathogenic
EXT1	194804	substitution	g. 118849385G>A	likely pathogenic
EXT1	194805	substitution	g. 118849385G>A	likely pathogenic
EXT1	194806	substitution	g. 118849385G>A	likely pathogenic
EXT1	194807	substitution	g. 118849385G>A	likely pathogenic
EXT1	194808	substitution	g. 118849385G>A	likely pathogenic
EXT1	194809	substitution	g. 118849385G>A	likely pathogenic
EXT1	194810	substitution	g. 118849385G>A	likely pathogenic
EXT1	194811	substitution	g. 118849385G>A	likely pathogenic
EXT1	194812	substitution	g. 118849385G>A	likely pathogenic
EXT1	194813	substitution	g. 118849385G>A	likely pathogenic
EXT1	194814	substitution	g. 118849385G>A	likely pathogenic
EXT1	194815	substitution	g. 118849384C>T	likely pathogenic
EXT1	194816	substitution	g. 118849384C>T	likely pathogenic
EXT1	194817	substitution	g. 118849384C>T	likely pathogenic



EXT1	194871	deletion	g. 118849346_118849369 del	likely pathogenic
EXT1	194872	substitution	g. 118849367T>C	likely pathogenic
EXT1	194873	substitution	g. 118849367T>C	likely pathogenic
EXT1	194874	substitution	g. 118849367T>C	likely pathogenic
EXT1	194875	substitution	g. 118849367T>C	likely pathogenic
EXT1	194876	substitution	g. 118849367T>A	likely pathogenic
EXT1	194877	substitution	g. 118849366C>G	likely pathogenic
EXT1	194878	substitution	g. 118849366C>A	likely pathogenic
EXT1	194879	substitution	g. 118849365T>A	likely pathogenic
EXT1	194880	substitution	g. 118849365T>A	likely pathogenic
EXT1	194881	substitution	g. 118849349G>A	likely pathogenic
EXT1	194882	substitution	g. 118849346C>T	likely pathogenic
EXT1	194883	substitution	g. 118849346C>T	likely pathogenic
EXT1	194884	substitution	g. 118849346C>T	likely pathogenic
EXT1	194885	substitution	g. 118849346C>T	likely pathogenic
EXT1	194886	substitution	g. 118849346C>T	likely pathogenic
EXT1	194887	substitution	g. 118849346C>T	likely pathogenic
EXT1	194888	duplication	g. 118849345dup	likely pathogenic
EXT1	194889	duplication	g. 118849345dup	likely pathogenic
EXT1	194890	substitution	g. 118849344T>G	VUS
EXT1	194891	substitution	g. 118847793G>C	VUS
EXT1	194892	substitution	g. 118847792T>C	VUS
EXT1	194893	substitution	g. 118847792T>C	VUS
EXT1	194894	substitution	g. 118847792T>A	VUS
EXT1	194895	substitution	g. 118847784A>G	likely pathogenic
EXT1	194896	substitution	g. 118847782G>A	benign
EXT1	194897	substitution	g. 118847781C>T	benign
EXT1	194898	deletion	g. 118847771_118847781 del	likely pathogenic
EXT1	194899	deletion	g. 118847779del	likely pathogenic
EXT1	194900	substitution	g. 118847755C>T	likely pathogenic
EXT1	194901	substitution	g. 118847755C>T	likely pathogenic
EXT1	194902	deletion	g. 118847756del	likely pathogenic
EXT1	194903	deletion	g. 118847751del	likely pathogenic
EXT1	194904	duplication	g. 118847746dup	likely pathogenic
EXT1	194905	substitution	g. 118847739C>G	likely pathogenic
EXT1	194906	substitution	g. 118847730T>C	likely pathogenic
EXT1	194907	indel	g. 118843573_118847729 delinsG	likely pathogenic
EXT1	194908	substitution	g. 118847726C>T	likely pathogenic
EXT1	194909	substitution	g. 118847725C>T	likely pathogenic
EXT1	194910	substitution	g. 118847725C>T	likely pathogenic
EXT1	194911	substitution	g. 118847721G>A	likely pathogenic
EXT1	194912	deletion	g. 118847695del	likely pathogenic
EXT1	194913	deletion	g. 118847695del	likely pathogenic
EXT1	194914	substitution	g. 118847690A>C	likely pathogenic
EXT1	194915	substitution	g. 118847687A>T	likely pathogenic
EXT1	194916	substitution	g. 118847685G>A	likely pathogenic
EXT1	194917	substitution	g. 118847685G>A	likely pathogenic
EXT1	194918	substitution	g. 118847685G>A	likely pathogenic
EXT1	194919	substitution	g. 118847685G>A	likely pathogenic
EXT1	194920	deletion	g. 118847683_118847684 del	likely pathogenic

EXT1	194921	duplication	g.118847683dup	likely pathogenic
EXT1	194922	substitution	g.118847682C>G	likely pathogenic
EXT1	194923	substitution	g.118847682C>A	likely pathogenic
EXT1	194924	insertion	g.118842587_118842588 insC	likely pathogenic
EXT1	194925	deletion	g.118842587del	likely pathogenic
EXT1	194926	deletion	g.118842583del	likely pathogenic
EXT1	194927	duplication	g.118842574_118842575 dup	likely pathogenic
EXT1	194928	substitution	g.118842561G>A	likely pathogenic
EXT1	194929	substitution	g.118842554T>C	likely pathogenic
EXT1	194930	duplication	g.118842555dup	likely pathogenic
EXT1	194931	substitution	g.118842540T>A	likely pathogenic
EXT1	194932	substitution	g.118842540T>A	likely pathogenic
EXT1	194933	substitution	g.118842540T>A	likely pathogenic
EXT1	194934	duplication	g.118842540dup	likely pathogenic
EXT1	194935	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194936	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194937	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194938	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194939	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194940	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194941	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194942	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194943	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194944	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194945	deletion	g.118842537_118842540 del	likely pathogenic
EXT1	194946	substitution	g.118842537G>A	likely pathogenic
EXT1	194947	substitution	g.118842537G>A	likely pathogenic
EXT1	194948	substitution	g.118842534G>A	likely pathogenic
EXT1	194949	substitution	g.118842534G>A	likely pathogenic
EXT1	194950	substitution	g.118842528G>A	likely pathogenic
EXT1	194951	substitution	g.118842528G>A	likely pathogenic
EXT1	194952	deletion	g.118842530_118842531 del	likely pathogenic
EXT1	194953	substitution	g.118842518C>T	likely pathogenic
EXT1	194954	substitution	g.118842518C>T	likely pathogenic
EXT1	194955	deletion	g.118842518del	likely pathogenic
EXT1	194956	substitution	g.118842516C>A	likely pathogenic
EXT1	194957	substitution	g.118842508A>C	likely pathogenic
EXT1	194958	substitution	g.118842482A>C	likely pathogenic
EXT1	194959	deletion	g.118842476_118842477 del	likely pathogenic

EXT1	194960	substitution	g.118842468C>A	likely pathogenic
EXT1	194961	substitution	g.118842468C>A	likely pathogenic
EXT1	194962	substitution	g.118842468C>A	likely pathogenic
EXT1	194963	substitution	g.118842467A>C	likely pathogenic
EXT1	194964	substitution	g.118842405G>T	benign
EXT1	194965	substitution	g.118834838T>G	likely pathogenic
EXT1	194966	deletion	g.118834836_118834839 del	likely pathogenic
EXT1	194967	substitution	g.118834837C>G	likely pathogenic
EXT1	194968	substitution	g.118834837C>G	likely pathogenic
EXT1	194969	substitution	g.118834837C>G	likely pathogenic
EXT1	194970	substitution	g.118834837C>G	likely pathogenic
EXT1	194971	substitution	g.118834830G>A	likely pathogenic
EXT1	194972	deletion	g.118834823_118834824 del	likely pathogenic
EXT1	194973	deletion	g.118834820del	likely pathogenic
EXT1	194974	substitution	g.118834814T>A	likely pathogenic
EXT1	194975	deletion	g.118834808_118834809 del	likely pathogenic
EXT1	194976	duplication	g.118834801dup	likely pathogenic
EXT1	194977	duplication	g.118834800dup	likely pathogenic
EXT1	194978	deletion	g.118834782_118834789 del	likely pathogenic
EXT1	194979	deletion	g.118834782_118834789 del	likely pathogenic
EXT1	194980	duplication	g.118834787dup	likely pathogenic
EXT1	194981	substitution	g.118834782T>A	likely pathogenic
EXT1	194982	duplication	g.118834779dup	likely pathogenic
EXT1	194983	substitution	g.118834761C>T	likely pathogenic
EXT1	194984	duplication	g.118834760dup	likely pathogenic
EXT1	194985	duplication	g.118834759dup	likely pathogenic
EXT1	194986	deletion	g.118834755del	likely pathogenic
EXT1	194987	deletion	g.118834753_118834754 del	likely pathogenic
EXT1	194988	deletion	g.118834749del	likely pathogenic
EXT1	194989	substitution	g.118834747A>C	likely pathogenic
EXT1	194990	substitution	g.118834745G>C	likely pathogenic
EXT1	194991	deletion	g.118834737del	likely pathogenic
EXT1	194992	deletion	g.118834729del	likely pathogenic
EXT1	194993	substitution	g.118834724G>C	likely pathogenic
EXT1	194994	deletion	g.118834719del	likely pathogenic
EXT1	194995	deletion	g.118834719del	likely pathogenic
EXT1	194996	substitution	g.118834717G>C	likely pathogenic
EXT1	194997	substitution	g.118834717G>C	likely pathogenic
EXT1	194998	deletion	g.118832034_118834713 del	likely pathogenic
EXT1	194999	deletion	g.118834711del	likely pathogenic
EXT1	195000	substitution	g.118834706A>T	likely pathogenic
EXT1	195001	substitution	g.118834704C>T	likely pathogenic
EXT1	195002	substitution	g.118834703C>T	likely pathogenic
EXT1	195003	substitution	g.118834703C>T	likely pathogenic
EXT1	195004	substitution	g.118834703C>A	likely pathogenic
EXT1	195005	duplication	g.118834702dup	likely pathogenic
EXT1	195006	duplication	g.118834702dup	likely pathogenic

EXT1	195007	substitution	g. 118834702A>C	likely pathogenic
EXT1	195008	deletion	g. 118834698_118834703 del	likely pathogenic
EXT1	195009	substitution	g. 118832035T>C	likely pathogenic
EXT1	195010	substitution	g. 118832035T>C	likely pathogenic
EXT1	195011	substitution	g. 118832035T>C	likely pathogenic
EXT1	195012	substitution	g. 118832034C>T	likely pathogenic
EXT1	195013	substitution	g. 118832030A>C	likely pathogenic
EXT1	195014	substitution	g. 118832028T>A	likely pathogenic
EXT1	195015	deletion	g. 118832026del	likely pathogenic
EXT1	195016	insertion	g. 118832020_118832021 insA	likely pathogenic
EXT1	195017	substitution	g. 118832020G>A	benign
EXT1	195018	deletion	g. 118832025del	likely pathogenic
EXT1	195019	duplication	g. 118832025dup	likely pathogenic
EXT1	195020	duplication	g. 118832025dup	likely pathogenic
EXT1	195021	duplication	g. 118832025dup	likely pathogenic
EXT1	195022	duplication	g. 118832025dup	likely pathogenic
EXT1	195023	duplication	g. 118832025dup	likely pathogenic
EXT1	195024	duplication	g. 118832025dup	likely pathogenic
EXT1	195025	duplication	g. 118832025dup	likely pathogenic
EXT1	195026	duplication	g. 118832025dup	likely pathogenic
EXT1	195027	duplication	g. 118832019dup	likely pathogenic
EXT1	195028	duplication	g. 118832019dup	likely pathogenic
EXT1	195029	duplication	g. 118832019dup	likely pathogenic
EXT1	195030	substitution	g. 118831994G>A	likely pathogenic
EXT1	195032	deletion	g. 118831988del	likely pathogenic
EXT1	195033	deletion	g. 118831988del	likely pathogenic
EXT1	195034	deletion	g. 118831988del	likely pathogenic
EXT1	195035	deletion	g. 118831988del	likely pathogenic
EXT1	195036	deletion	g. 118831988del	likely pathogenic
EXT1	195037	deletion	g. 118831988del	likely pathogenic
EXT1	195038	deletion	g. 118831988del	likely pathogenic
EXT1	195039	duplication	g. 118831988dup	likely pathogenic
EXT1	195040	duplication	g. 118831988dup	likely pathogenic
EXT1	195041	duplication	g. 118831988dup	likely pathogenic
EXT1	195042	duplication	g. 118831988dup	likely pathogenic
EXT1	195043	duplication	g. 118831988dup	likely pathogenic
EXT1	195044	duplication	g. 118831988dup	likely pathogenic
EXT1	195045	duplication	g. 118831988dup	likely pathogenic
EXT1	195046	duplication	g. 118831988dup	likely pathogenic
EXT1	195047	duplication	g. 118831988dup	likely pathogenic
EXT1	195048	duplication	g. 118831988dup	likely pathogenic
EXT1	195049	deletion	g. 118831982del	likely pathogenic
EXT1	195050	deletion	g. 118831982del	likely pathogenic
EXT1	195051	deletion	g. 118831982del	likely pathogenic
EXT1	195052	deletion	g. 118831982del	likely pathogenic
EXT1	195053	deletion	g. 118831982del	likely pathogenic
EXT1	195054	deletion	g. 118831982del	likely pathogenic
EXT1	195055	deletion	g. 118831982del	likely pathogenic
EXT1	195056	deletion	g. 118831982del	likely pathogenic
EXT1	195057	deletion	g. 118831982del	likely pathogenic
EXT1	195058	deletion	g. 118831982del	likely pathogenic
EXT1	195059	deletion	g. 118831982del	likely pathogenic

EXT1	195060	deletion	g. 118831982del	likely pathogenic
EXT1	195061	deletion	g. 118831982del	likely pathogenic
EXT1	195062	deletion	g. 118831982del	likely pathogenic
EXT1	195063	deletion	g. 118831982del	likely pathogenic
EXT1	195064	deletion	g. 118831982del	likely pathogenic
EXT1	195065	deletion	g. 118831982del	likely pathogenic
EXT1	195066	deletion	g. 118831982del	likely pathogenic
EXT1	195067	deletion	g. 118831982del	likely pathogenic
EXT1	195068	deletion	g. 118831982del	likely pathogenic
EXT1	195069	deletion	g. 118831982del	likely pathogenic
EXT1	195070	deletion	g. 118831982del	likely pathogenic
EXT1	195071	deletion	g. 118831982del	likely pathogenic
EXT1	195072	deletion	g. 118831982del	likely pathogenic
EXT1	195073	deletion	g. 118831982del	likely pathogenic
EXT1	195074	deletion	g. 118831982del	likely pathogenic
EXT1	195075	deletion	g. 118831982del	likely pathogenic
EXT1	195076	deletion	g. 118831982del	likely pathogenic
EXT1	195077	deletion	g. 118831982del	likely pathogenic
EXT1	195078	deletion	g. 118831982del	likely pathogenic
EXT1	195079	deletion	g. 118831982del	likely pathogenic
EXT1	195080	deletion	g. 118831982del	likely pathogenic
EXT1	195081	deletion	g. 118831982del	likely pathogenic
EXT1	195082	deletion	g. 118831982del	likely pathogenic
EXT1	195083	deletion	g. 118831982del	likely pathogenic
EXT1	195084	deletion	g. 118831982del	likely pathogenic
EXT1	195085	deletion	g. 118831982del	likely pathogenic
EXT1	195086	deletion	g. 118831978_118831979 del	likely pathogenic
EXT1	195087	deletion	g. 118831975del	likely pathogenic
EXT1	195088	deletion	g. 118831978_118831979 del	likely pathogenic
EXT1	195089	substitution	g. 118831974G>A	likely pathogenic
EXT1	195090	substitution	g. 118831968G>A	likely pathogenic
EXT1	195091	substitution	g. 118831964G>A	likely pathogenic
EXT1	195092	deletion	g. 118831957del	likely pathogenic
EXT1	195093	duplication	g. 118831942dup	likely pathogenic
EXT1	195094	substitution	g. 118831929G>A	likely pathogenic
EXT1	195095	substitution	g. 118831924G>C	likely pathogenic
EXT1	195096	substitution	g. 118831917G>A	likely pathogenic
EXT1	195097	deletion	g. 118831919del	likely pathogenic
EXT1	195098	substitution	g. 118831914C>T	likely pathogenic
EXT1	195099	substitution	g. 118831914C>G	likely pathogenic
EXT1	195100	substitution	g. 118831914C>A	likely pathogenic
EXT1	195101	substitution	g. 118831914C>A	likely pathogenic
EXT1	195102	substitution	g. 118831914C>A	likely pathogenic
EXT1	195103	substitution	g. 118831908C>T	likely pathogenic
EXT1	195104	substitution	g. 118830771T>C	likely pathogenic
EXT1	195105	substitution	g. 118830770C>G	likely pathogenic
EXT1	195106	substitution	g. 118830770C>A	likely pathogenic
EXT1	195107	substitution	g. 118830770C>A	likely pathogenic
EXT1	195108	deletion	g. (118819617_11882511 0)_(118830770_1188319 14)del	likely pathogenic
EXT1	195109	substitution	g. 118830756C>T	likely pathogenic

EXT1	195110	duplication	g.118830756dup	likely pathogenic
EXT1	195111	substitution	g.118830755C>T	likely pathogenic
EXT1	195112	substitution	g.118830749A>T	likely pathogenic
EXT1	195113	deletion	g.118830738del	likely pathogenic
EXT1	195114	deletion	g.118830738del	likely pathogenic
EXT1	195115	duplication	g.118830738dup	likely pathogenic
EXT1	195116	deletion	g.118830723_118830736 del	likely pathogenic
EXT1	195117	substitution	g.118830719C>T	likely pathogenic
EXT1	195118	substitution	g.118830719C>T	likely pathogenic
EXT1	195119	substitution	g.118830719C>T	likely pathogenic
EXT1	195120	deletion	g.118830710del	likely pathogenic
EXT1	195121	substitution	g.118830697C>T	likely pathogenic
EXT1	195122	deletion	g.118830687del	likely pathogenic
EXT1	195123	deletion	g.118830665_118830688 del	likely pathogenic
EXT1	195124	substitution	g.118830682C>A	likely pathogenic
EXT1	195125	substitution	g.118830673C>G	VUS
EXT1	195126	substitution	g.118825226G>T	VUS
EXT1	195127	substitution	g.118825202T>C	likely pathogenic
EXT1	195128	substitution	g.118825202T>C	likely pathogenic
EXT1	195129	deletion	g.118825202del	likely pathogenic
EXT1	195130	substitution	g.118825201C>G	likely pathogenic
EXT1	195131	insertion	g.118825199_118825200 insGGAAA	likely pathogenic
EXT1	195132	deletion	g.118825191del	likely pathogenic
EXT1	195133	deletion	g.118825191del	likely pathogenic
EXT1	195134	deletion	g.118825191del	likely pathogenic
EXT1	195135	deletion	g.118825191del	likely pathogenic
EXT1	195136	deletion	g.118825189del	likely pathogenic
EXT1	195137	deletion	g.118825189del	likely pathogenic
EXT1	195138	deletion	g.118825189del	likely pathogenic
EXT1	195139	deletion	g.118825185_118825186 del	likely pathogenic
EXT1	195140	indel	g.118825170_118825176 delins13	likely pathogenic
EXT1	195141	substitution	g.118825174G>T	likely pathogenic
EXT1	195142	substitution	g.118825174G>T	likely pathogenic
EXT1	195143	deletion	g.118825164_118825173 del	likely pathogenic
EXT1	195144	duplication	g.118825170dup	likely pathogenic
EXT1	195145	indel	g.118825152_118825156 delinsTCA	likely pathogenic
EXT1	195146	duplication	g.118825154dup	likely pathogenic
EXT1	195147	substitution	g.118825148A>G	likely pathogenic
EXT1	195148	substitution	g.118825148A>G	likely pathogenic
EXT1	195149	substitution	g.118825148A>G	likely pathogenic
EXT1	195150	substitution	g.118825148A>G	likely pathogenic
EXT1	195151	substitution	g.118825148A>C	likely pathogenic
EXT1	195152	substitution	g.118825137C>A	likely pathogenic
EXT1	195153	substitution	g.118825137C>A	likely pathogenic
EXT1	195154	insertion	g.118825130_118825131 ins5	likely pathogenic
EXT1	195155	deletion	g.118825120del	likely pathogenic

EXT1	195156	deletion	g.118825120del	likely pathogenic
EXT1	195157	substitution	g.118825113C>A	likely pathogenic
EXT1	195158	substitution	g.118825110C>T	likely pathogenic
EXT1	195159	substitution	g.118825110C>G	likely pathogenic
EXT1	195160	substitution	g.118825110C>A	likely pathogenic
EXT1	195161	substitution	g.118825110C>A	likely pathogenic
EXT1	195162	duplication	g.118825109dup	likely pathogenic
EXT1	195163	substitution	g.118825109A>G	likely pathogenic
EXT1	195164	substitution	g.118825109A>G	likely pathogenic
EXT1	195165	substitution	g.118819618T>G	likely pathogenic
EXT1	195166	substitution	g.118819617C>T	likely pathogenic
EXT1	195167	substitution	g.118819617C>G	likely pathogenic
EXT1	195168	substitution	g.118819616C>G	likely pathogenic
EXT1	195169	deletion	g.118819595_118819598 del	likely pathogenic
EXT1	195170	substitution	g.118819594C>T	likely pathogenic
EXT1	195171	substitution	g.118819594C>T	likely pathogenic
EXT1	195172	substitution	g.118819594C>T	likely pathogenic
EXT1	195173	substitution	g.118819593C>T	likely pathogenic
EXT1	195174	substitution	g.118819593C>T	likely pathogenic
EXT1	195175	deletion	g.118819581del	likely pathogenic
EXT1	195176	substitution	g.118819578C>T	benign
EXT1	195177	deletion	g.118819567_118819571 del	likely pathogenic
EXT1	195178	deletion	g.118819569del	likely pathogenic
EXT1	195179	deletion	g.118819569del	likely pathogenic
EXT1	195180	substitution	g.118819563G>T	likely pathogenic
EXT1	195181	substitution	g.118819563G>T	likely pathogenic
EXT1	195182	substitution	g.118819563G>T	likely pathogenic
EXT1	195183	substitution	g.118819560G>A	VUS
EXT1	195184	deletion	g.118819559_118819560 del	likely pathogenic
EXT1	195185	deletion	g.118819559_118819560 del	likely pathogenic
EXT1	195186	substitution	g.118819543C>T	likely pathogenic
EXT1	195187	substitution	g.118819542C>T	likely pathogenic
EXT1	195188	substitution	g.118819542C>T	likely pathogenic
EXT1	195189	substitution	g.118819542C>T	likely pathogenic
EXT1	195190	substitution	g.118819532T>A	likely pathogenic
EXT1	195191	substitution	g.118819529C>A	likely pathogenic
EXT1	195192	deletion	g.118819525del	likely pathogenic
EXT1	195193	deletion	g.118819523del	likely pathogenic
EXT1	195194	substitution	g.118819522C>T	likely pathogenic
EXT1	195195	substitution	g.118819522C>T	VUS
EXT1	195196	substitution	g.118819521C>T	likely pathogenic
EXT1	195197	substitution	g.118819520C>A	likely pathogenic
EXT1	195198	substitution	g.118819504C>T	likely pathogenic
EXT1	195199	substitution	g.118819504C>T	likely pathogenic
EXT1	195200	substitution	g.118819503C>T	likely pathogenic
EXT1	195201	substitution	g.118819497G>A	VUS
EXT1	195202	deletion	g.118819446_118819481 del	likely pathogenic
EXT1	195203	substitution	g.118819475C>A	likely pathogenic
EXT1	195204	deletion	g.118819475del	likely pathogenic

EXT1	195205	deletion	g.118819475del	likely pathogenic
EXT1	195206	deletion	g.118819460_118819462del	likely pathogenic
EXT1	195207	substitution	g.118819455C>G	likely pathogenic
EXT1	195208	substitution	g.118819455C>G	likely pathogenic
EXT1	195209	substitution	g.118819454A>C	likely pathogenic
EXT1	195210	substitution	g.118819454A>C	likely pathogenic
EXT1	195211	substitution	g.118819454A>C	likely pathogenic
EXT1	195212	duplication	g.118817122dup	likely pathogenic
EXT1	195213	substitution	g.118817120G>T	likely pathogenic
EXT1	195214	substitution	g.118817120G>C	likely pathogenic
EXT1	195215	deletion	g.118817120del	likely pathogenic
EXT1	195216	deletion	g.118817112del	likely pathogenic
EXT1	195217	duplication	g.118817102_118817105dup	likely pathogenic
EXT1	195218	duplication	g.118817102_118817105dup	likely pathogenic
EXT1	195219	deletion	g.118817099_118817102del	likely pathogenic
EXT1	195220	duplication	g.118817099_118817102dup	likely pathogenic
EXT1	195221	substitution	g.118817074G>A	likely pathogenic
EXT1	195222	deletion	g.118817068_118817075del	likely pathogenic
EXT1	195223	deletion	g.118817036del	likely pathogenic
EXT1	195224	deletion	g.118817034del	likely pathogenic
EXT1	195225	substitution	g.118816996G>A	likely pathogenic
EXT1	195226	substitution	g.118816996G>A	likely pathogenic
EXT1	195227	substitution	g.118816996G>A	likely pathogenic
EXT1	195228	deletion	g.118816995del	likely pathogenic
EXT1	195229	substitution	g.118816993T>A	likely pathogenic
EXT1	195230	substitution	g.118816987G>A	likely pathogenic
EXT1	195231	substitution	g.118816982A>C	likely pathogenic
EXT1	195232	substitution	g.118816978C>A	likely pathogenic
EXT1	195233	indel	g.118816962_118816966delins13	likely pathogenic
EXT1	195234	deletion	g.118816967del	likely pathogenic
EXT1	195235	substitution	g.118816963G>A	likely pathogenic
EXT1	195236	substitution	g.118816963G>A	likely pathogenic
EXT1	195237	substitution	g.118816963G>A	likely pathogenic
EXT1	195238	substitution	g.118812117C>T	likely pathogenic
EXT1	195239	deletion	g.118812097_118812109del	likely pathogenic
EXT1	195240	deletion	g.118812099_118812100del	likely pathogenic
EXT1	195241	substitution	g.118812094G>A	likely pathogenic
EXT1	195242	substitution	g.118812094G>A	likely pathogenic
EXT1	195243	substitution	g.118812091G>A	likely pathogenic
EXT1	195244	substitution	g.118812091G>A	likely pathogenic
EXT1	195245	substitution	g.118812088G>A	likely pathogenic
EXT1	195246	deletion	g.118812072del	likely pathogenic
EXT1	195247	deletion	g.118812049_118812059del	likely pathogenic
EXT1	195248	deletion	g.118811817del	VUS

EXT1	223729		g. (118811950_119123286)_qterdelins[NC_000004.11:(107700001_114100000)_qter]	likely pathogenic
EXT1	223730		g. (119038109-119106577)_qterdelins[NC_000019.9:(35762292_35940346)_qter]	likely pathogenic
EXT1	255529	deletion	g.118831982del	pathogenic
EXT1	267922	substitution	g.118819578C>T	benign
EXT1	268930	substitution	g.119123172T>C	benign
EXT1	268931	substitution	g.119123169T>C	benign
EXT1	268932	substitution	g.118812044C>T	benign
EXT1	268933	substitution	g.119122886T>C	likely benign
EXT1	268934	deletion	g.119122315_119122318del	likely benign
EXT1	276429	substitution	g.118849346C>T	pathogenic
EXT1	276430	substitution	g.118834762G>A	likely benign
EXT1	276431	substitution	g.118832034C>A	pathogenic
EXT1	276433	substitution	g.118830770C>G	pathogenic
EXT1	276434	deletion	g.118825136_118825137del	pathogenic
EXT1	276435	deletion	g.118825129del	pathogenic
EXT1	276436	substitution	g.118825110C>A	pathogenic
EXT1	276437	deletion	g.118819569del	pathogenic
EXT1	276438	substitution	g.118817133C>G	pathogenic
EXT1	276439	deletion	g.119123082_119123083del	pathogenic
EXT1	276440	deletion	g.118812045_118812063del	pathogenic
EXT1	276441	substitution	g.119122830G>T	pathogenic
EXT1	276443	deletion	g.119122608del	pathogenic
EXT1	276444	deletion	g.119122569_119122583del	pathogenic
EXT1	276446	deletion	g.119122492_119122499del	pathogenic
EXT1	276447	deletion	g.119122322_119122325del	pathogenic
EXT1	345384	substitution	g.118847739C>G	VUS
EXT1	369936	deletion	g.118834704del	likely pathogenic
EXT1	439899	deletion	g.119123215_119123222del	VUS
EXT1	439900	deletion	g.119123114del	VUS
EXT1	439901	deletion	g.119123114del	VUS
EXT1	439902	deletion	g.119123114del	VUS
EXT1	439903	substitution	g.119123036G>A	VUS
EXT1	439904	substitution	g.119123036G>A	VUS
EXT1	439905	substitution	g.119123036G>A	VUS
EXT1	439906	duplication	g.119123024dup	VUS
EXT1	439907	deletion	g.119122986del	VUS
EXT1	439908	substitution	g.119122935G>C	VUS
EXT1	439909	substitution	g.119122935G>C	VUS
EXT1	439910	deletion	g.119122849del	VUS
EXT1	439911	substitution	g.119122786G>T	VUS

EXT1	439912	substitution	g. 119122734C>T	VUS
EXT1	439913	deletion	g. 119123226_119123230 del	VUS
EXT1	439914	deletion	g. 119123226_119123230 del	VUS
EXT1	439915	substitution	g. 119122686C>T	VUS
EXT1	439916	deletion	g. 119122666del	VUS
EXT1	439917	deletion	g. 119122666del	VUS
EXT1	439918	substitution	g. 119122649G>A	VUS
EXT1	439919	deletion	g. 119122544del	VUS
EXT1	439920	deletion	g. 119122538del	VUS
EXT1	439923	substitution	g. 119122478T>A	VUS
EXT1	439924	substitution	g. 119122448T>C	VUS
EXT1	439925	deletion	g. 119122345_119122346 del	VUS
EXT1	439926	deletion	g. (?_118811950)_(1188 49441_119122323)del	VUS
EXT1	439927	substitution	g. 118849437A>C	VUS
EXT1	439928	substitution	g. 118849385G>A	VUS
EXT1	439929	substitution	g. 118849384C>T	VUS
EXT1	439930	substitution	g. 118849384C>T	VUS
EXT1	439931	substitution	g. 118849384C>T	VUS
EXT1	439932	substitution	g. 118849384C>T	VUS
EXT1	439933	substitution	g. 118849372G>A	VUS
EXT1	439934	substitution	g. 118847783C>A	VUS
EXT1	439935	substitution	g. 118847782G>T	pathogenic
EXT1	439936	substitution	g. 118847782G>T	pathogenic
EXT1	439937	substitution	g. 118847756C>T	VUS
EXT1	439938	substitution	g. 118847756C>T	VUS
EXT1	439939	deletion	g. 118847688del	VUS
EXT1	439940	substitution	g. 118847685G>A	VUS
EXT1	439941	substitution	g. 118847682C>T	VUS
EXT1	439942	substitution	g. 118842540T>A	VUS
EXT1	439943	deletion	g. 118842476_118842477 del	pathogenic
EXT1	439944	substitution	g. 118842468C>A	likely pathogenic (dominant)
EXT1	439945	substitution	g. 118842468C>T	VUS
EXT1	439946	substitution	g. 118834838T>C	VUS
EXT1	439947	substitution	g. 118834805G>C	VUS
EXT1	439948	substitution	g. 118834786C>T	VUS
EXT1	439949	substitution	g. 118834786C>T	VUS
EXT1	439950	deletion	g. 118834724_118834727 del	VUS
EXT1	439951	substitution	g. 118832035T>C	VUS
EXT1	439952	deletion	g. 118832026del	VUS
EXT1	439953	deletion	g. 118832026del	VUS
EXT1	439954	deletion	g. 118831982del	VUS
EXT1	439955	substitution	g. 118831968G>A	VUS
EXT1	439956	deletion	g. 118831961del	VUS
EXT1	439957	substitution	g. 118831929G>A	VUS
EXT1	439958	substitution	g. 118831914C>A	VUS
EXT1	439959	substitution	g. 118830770C>A	VUS
EXT1	439960	substitution	g. 118830771T>A	VUS
EXT1	439961	substitution	g. 118825202T>A	VUS

EXT1	439962	deletion	g.118825191del	VUS
EXT1	439963	deletion	g.118825120del	VUS
EXT1	439964	deletion	g.118825120del	VUS
EXT1	439965	substitution	g.118819593C>T	VUS
EXT1	439966	deletion	g.118819581_118819582 del	VUS
EXT1	439967	substitution	g.118819580C>A	VUS
EXT1	439968	substitution	g.118819542C>T	VUS
EXT1	439969	deletion	g.118819499del	VUS
EXT1	439970	insertion	g.118817094_118817095 insGGCT	VUS
EXT1	439971	substitution	g.118817074G>A	VUS
EXT1	439972	duplication	g.118812123dup	VUS
EXT1	439973	substitution	g.118812094G>A	VUS
EXT1	439974	deletion	g.118811950_119123285 del	VUS
EXT1	472715	substitution	g.118847718C>T	VUS
EXT1	533679	substitution	g.118812019C>T	VUS
EXT1	533680	substitution	g.118825159T>C	likely benign
EXT1	533681	substitution	g.118825174G>A	likely benign
EXT1	533682	deletion	g.118831978_118831979 del	pathogenic
EXT1	533683	duplication	g.118831988dup	pathogenic
EXT1	533684	deletion	g.119122315_119122318 del	likely benign
EXT1	533685	indel	g.119122463_119122468 delinsGCCTT	pathogenic
EXT1	533686	duplication	g.119122546_119122547 dup	pathogenic
EXT1	533687	duplication	g.119122663_119122667 dup	pathogenic
EXT1	533688	substitution	g.119122868C>T	likely benign
EXT1	533689	deletion	g.119123227_119123230 del	pathogenic
EXT1	611421	substitution	g.119123281T>C	VUS
EXT1	621986	substitution	g.118830697C>T	likely benign
EXT1	621987	substitution	g.118847781C>T	likely benign
EXT1	659581	substitution	g.118825110C>A	pathogenic
EXT1	682779	substitution	g.118819578C>T	benign
EXT1	682780	substitution	g.118817114G>T	pathogenic (dominant)
EXT1	682781	substitution	g.118847782G>A	benign
EXT1	683272	deletion	g.118842494del	likely pathogenic
EXT1	690147	substitution	g.119123178C>T	likely benign
EXT1	721775	substitution	g.119122949C>T	likely benign
EXT1	721776	substitution	g.119123278G>A	VUS
EXT1	729931	substitution	g.118812060C>T	likely pathogenic (dominant)
EXT1	731295	substitution	g.118849388C>T	pathogenic (dominant)
EXT1	731297	deletion	g.118819602_118819603 del	pathogenic (dominant)
EXT1	731298	substitution	g.118825199A>T	pathogenic (dominant)
EXT1	795163	substitution	g.119122329A>C	likely pathogenic